Perspectives in

MEDICAL AND GENETIC RESEARCH

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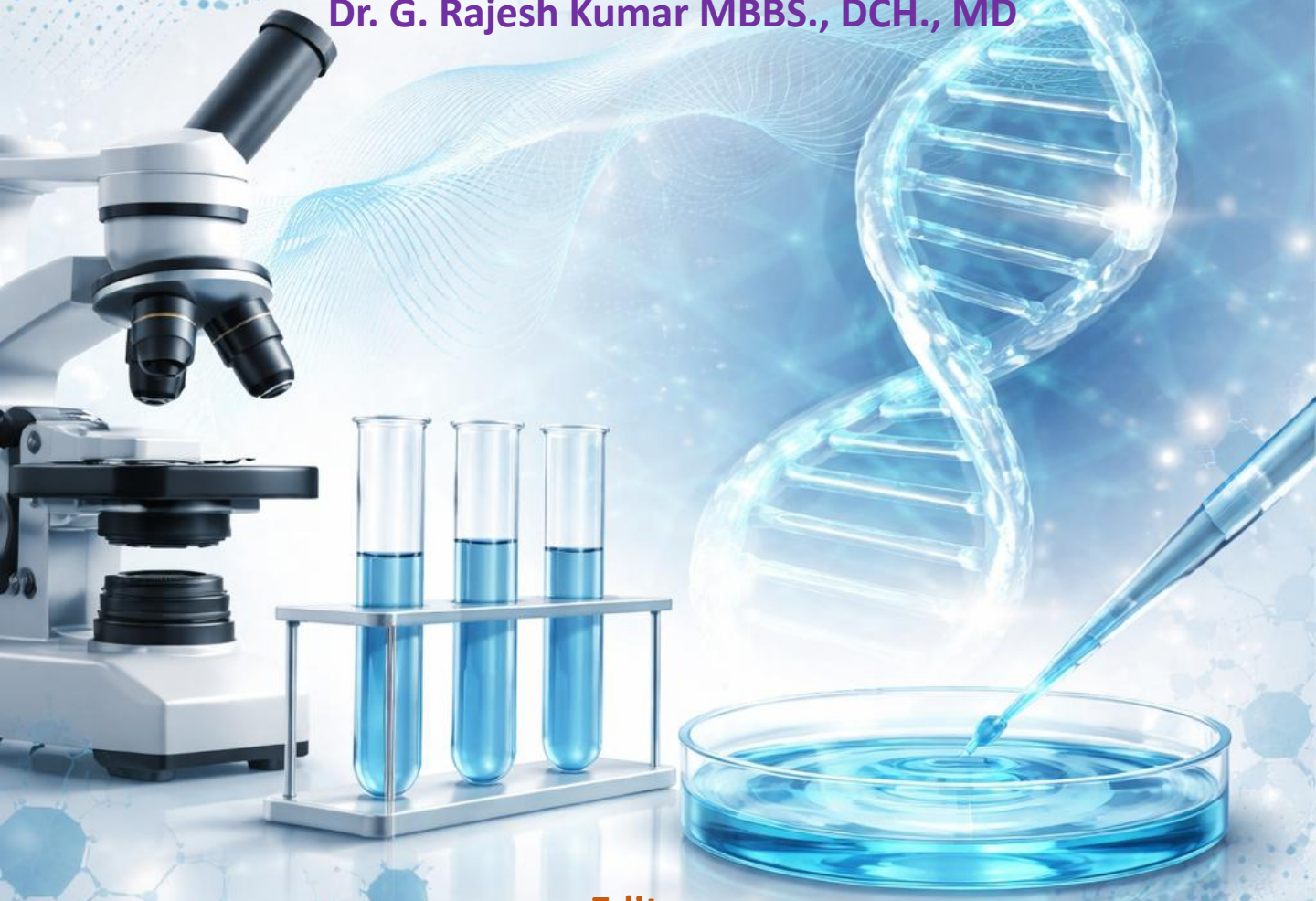

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Perspectives in Medical and Genetic Research

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PREFACE

The domain of medical and genetic research is constantly changing due to the persistent improvement of technology and an increasing knowledge of human biology. Perspectives in Medical and Genetic Research is a collaborative effort to provide a comprehensive and insightful overview of some of the major topics that influence modern medicine and science. The book is a compilation of a variety of different chapters that discuss a number of different diseases and conditions, ranging from common diseases to rare diseases such as genetic disorders, cancer biology, neurological disorders, metabolic syndromes, and new forms of therapy. From basic overviews of diseases such as Crohn's disease, Alzheimer's disease, and Polycystic Ovary Syndrome (PCOS), to in-depth overviews of rare diseases such as Moyamoya disease and methemoglobinemia, this book provides a comprehensive overview of a number of different diseases and conditions.

A major emphasis has been given to the importance of the role played by genetics and bioinformatics in the field. Chapters on the role of mutations in the CFTR gene, protein prediction in oral cancer, analysis of mutations in the genes causing progeria, and the application of bioinformatics show the impact of these tools on the understanding of complex biological processes. Furthermore, topics such as gene therapy and DNA fingerprinting show the expanding horizons of the field. Studies on diseases such as leukemia, pancreatic cancer, lung cancer, and spinal tumors show the continued efforts being made to improve treatment and patient outcome. Discussions on congenital defects, malnutrition, alopecia, and blood groups show the holistic view of health and disease.

It is the hope of the editors that this book will prove to be an important source of reference for students, researchers, and professionals in the fields of medicine, genetics, and life sciences. The book is also hoped to inspire readers to think critically and conduct research on these topics and to develop an appreciation for the complexities involved in these fields of study. The editors would like to express their sincere gratitude for the constant support and encouragement extended by the Secretary, Principal, staff members, and students, whose contributions and cooperation have been invaluable in the successful completion of this work. It is our earnest hope that Perspectives in Medical and Genetic Research will prove to be a significant contribution to the field of academic studies and research and will inspire readers to explore new frontiers in the fields of medical and genetic research.

- Dr. M. Thenmozhi

Dear Students, Parents, and Well-Wishers

It is a matter of immense satisfaction to witness our Grade XII students presenting this book on Perspectives in Medical and Genetic Research. This initiative reflects the progressive academic environment we aim to cultivate — one that encourages inquiry, research, and responsible learning.

In today's world, scientific awareness is no longer optional; it is essential. Understanding genetic disorders helps build a society that is informed, compassionate, and supportive of medical advancement. When students engage with such meaningful topics, they move beyond examination preparation and step into the realm of real-world understanding.

At Sree Narayana Mission Senior Secondary School, our commitment has always been to provide holistic education — nurturing intellectual growth along with ethical values. Projects like this strengthen students' confidence, research capabilities, and sense of social responsibility.

I appreciate the efforts of the Principal and the Teachers who have guided the students in bringing out this publication. My congratulations to the young contributors for their dedication and scholarly spirit.

May this endeavour mark the beginning of many such academic milestones and inspire our students to pursue excellence in every sphere of life.

With blessings and best wishes,

Deepak Devarajan

Secretary

Sree Narayana Mission Senior Secondary School

Chennai

Dear Students, Teachers, and Readers

It gives me immense pride and happiness to present this book on Perspectives in Medical and Genetic Research, thoughtfully prepared by our Grade XII students. This work reflects not only academic excellence but also scientific curiosity, critical thinking, and a commitment to understanding the complexities of human life.

Genetics is one of the most fascinating and rapidly advancing branches of science. From understanding inherited disorders to exploring revolutionary technologies such as gene therapy, this field has transformed modern medicine. By studying genetic diseases, our students are not merely learning about conditions and chromosomes - they are learning empathy, responsibility, and the importance of scientific research in improving lives.

This e-book is a testament to the dedication of our students and the guidance of our teachers. It demonstrates their ability to research, analyze, and present scientific concepts in a clear and meaningful manner. I am confident that this publication will serve as a valuable resource for peers and inspire young minds to pursue careers in medicine, biotechnology, and research.

At our institution, we believe education must go beyond textbooks. It must encourage inquiry, innovation, and social awareness. This initiative beautifully embodies that vision.

I congratulate our Grade XII students for their commendable effort and appreciate the teachers who mentored them throughout this project. May this be the beginning of many more scholarly contributions from our young scientists.

Wishing all our students continued success in their academic journey and beyond.

With best wishes,

Mrs. Jayalakshmi S

Principal

Sree Narayana Mission Senior Secondary School

Chennai

INDEX

Chapter - 1

A Review on Basic Characteristics of Crohn's Disease

Dakshita Sai S

ABSTRACT:

Crohn's disease is a long-term inflammatory bowel condition that affects the colon or ileum. Any other part of the body, from the mouth to the anus, may likewise be impacted. The physiological function of the epithelial barrier is compromised in Crohn's disease due to several factors, such as environmental influences, genetic predisposition, and gut flora. The symptoms are extremely bothersome and lead to a significant decline in quality of life, sometimes resulting in debilitating permanent damage to the digestive system that necessitates lifelong enteral or parenteral nutrition. To achieve a correct and precise diagnosis, a suitably chosen diagnostic approach for a specific clinical entity is essential. Common diagnostic methods consist of lab tests, histopathological evaluation, endoscopy, X-rays, CT scans, ultrasound imaging, and MRI. The study of medical biology and metalloproteinase analysis has also been useful in identifying alterations resulting from Crohn's disease. This section presents a comprehensive overview of the most recent findings related to Crohn's disease, including its genetic aspects, symptoms, morphology, diagnosis, and treatment options

Keywords: Crohn's disease; characteristics, symptoms, diagnostics, Treatment

INTRODUCTION:

Crohn's disease is an inflammatory bowel condition that causes swelling and irritation of tissues in the digestive tract. This could lead to stomach pain, severe diarrhea, fatigue, loss of weight, and nutritional shortages. Crohn's disease was identified in 1932 by three physicians - Burrill Crohn, Leon Ginzberg, and Gordon B Oppenheimer[1]. There isn't a definitive first victim of Crohn's disease; historical accounts of similar ailments indicate that the condition has existed for many years. Inflammation from Crohn's disease can influence various regions of the gastrointestinal tract. Crohn's disease typically impacts the final section and the initial portion of the small and large intestine. The inflammation regularly lengthens in the deeper layers of the both intestine. Crohn's disease can be both distressing and weakening (to render someone's body or mind less strong). At times, it can result in severe or life-threatening issues. While a definitive remedy for Crohn's disease (Figure 1.1) remains elusive, treatments can significantly relieve symptoms and potentially lead to long-lasting remission and reduction of inflammation[2,3].

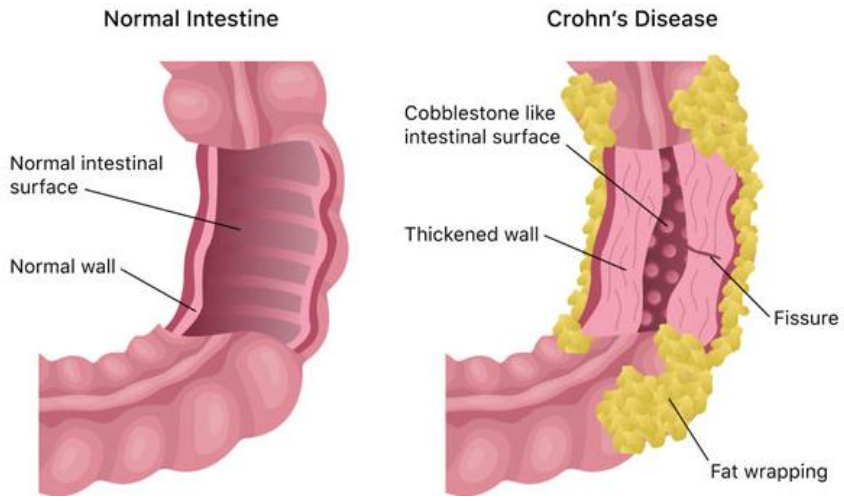

Figure 1.1: Comparison between the normal and crohn's disease intestine

TYPES OF CROHNS DISEASE[4]:

Chronic illness can impact any section of digestive system, ranging from mouth to anus (butt-hole). It usually leads to inflammation in small and/or large bowel (Figure 1.2).

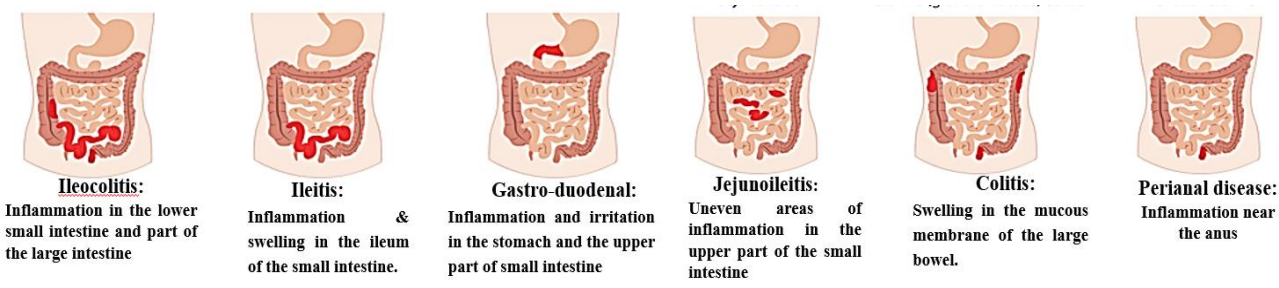

Figure 1.2: Types of Crohns Disease

CAUSES OF CROHNS DISEASE[4]:

The exact origin of Crohn's disease is mysterious, but it is associated with an abnormal immune response.

- Typically, the immune system shifts into combat mode to eliminate germs that invade the body. Inflammation indicates that body is combating the pathogens. After the danger is removed, the immune system settles down and the swelling fades away.
- An autoimmune reaction occurs when the immune system becomes hyperactive and begins to assault the body's own cells. The immune system might mistakenly target

bacteria that are typically present in the gut, leading to inflammation in the intestines associated with Crohn's disease.

- Crohn's can occasionally be hereditary. It may be that the factors triggering the immune system to harm the body's healthy cells are inherited.

RISK FACTORS[5]:

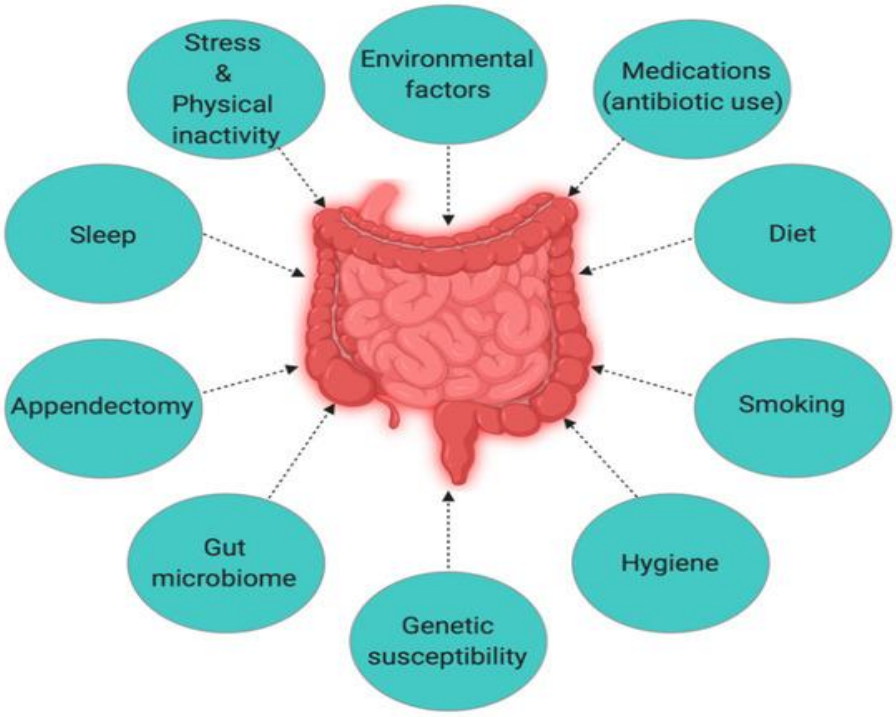

Figure 1.3: Risk Factors

COMPLICATIONS[7]:

- *Abscesses*: Infectious pus-filled sacs that develop in the digestive system or abdominal region. Formation of pockets filled with pus
- *Anal fissures:* Minor lacerations in the anus that lead to bleeding, discomfort and itching.
- *Fistulas:* A tunnel-like opening in the abdomen that links 2 parts of the body, resulting in a connection between the rectum or anal canal within the body and the skin around the anus outside the body.
- *Intestinal blockage:* A complete or partial obstruction in the bowel. Obstructions may arise when scar tissue develops or when narrowing of the abdomen, known as strictures, takes place.
- *Malnutrition:* Insufficient intake of essential nutrients. Inflammation may hinder body's ability to absorb necessary nutrients. Signs can hinder individual's ability to eat the essential food for energy. Deprivation and Lack of Nutrition

➢ *Anemia:* Reduced levels of blood cells. About 1 in 3 individuals with Crohn's disease experience anemia.

SYMPTOMS OF CROHN'S DISEASE[6]:

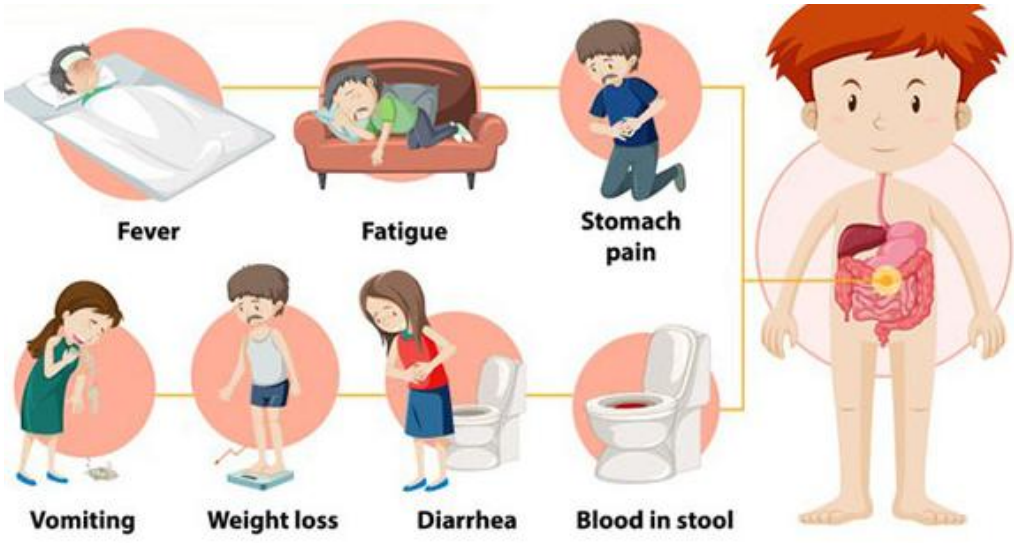

Figure 1.4: Symptoms of Crohn's Disease

DIAGNOSIS[8]:

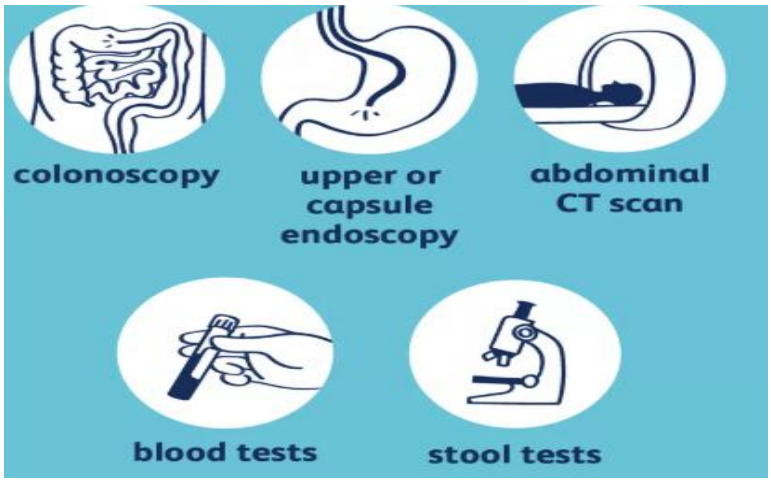

Figure 1.4: Diagnosis

TREATMENTS[9]:

At present, there is no remedy for Crohn's disease, and no single treatment is effective for all individuals. Nonetheless, multiple medications have received approval for treating Crohn's disease. A primary objective of medical therapy is to lessen the inflammation that causes

symptoms. A different objective is to enhance long-term outcomes by reducing complications. In optimal situations, this could result in not just alleviation of symptoms but also sustained remission.

- *Medicines:* Management of Crohn's disease involves medications that decrease inflammation and soothe of patient immune reactions. Research indicates that addressing the inflammation probably enhances the lasting results of Crohn's disease. Patient might also require therapies to address particular symptoms, such as pain and diarrhea.
- *Nutrition:* Obtaining proper nutrition can enhance the health and occasionally address the illness in the short term. If the patient are not able to obtain the necessary nutrition, patient might require formula administered via a feeding tube (enteral nutrition). Based on patient disease's condition, medical advisor will frequently provide tailored recommendations and conduct tests to make sure patient nutritional requirements are fulfilled.
- *Operation:* Surgery may address complications arising from Crohn's disease. Surgery might be required to eliminate intestinal perforations (holes), fistulas, strictures, and obstructions.

PREVENTION[9]:

- *Ceasing smoking:*

  Giving up smoking is the most effective action to lower the chances of flare-ups and complications.
- *Steering clear of medications that may induce flare-ups:*

  Using specific drugs can elevate the likelihood of flare-ups. This encompasses certain varieties of NSAIDs (non-steroidal anti-inflammatory drugs).
- *Steering clear of foods that could cause undesirable symptoms:*

  Lactose intolerance necessitates staying away from dairy products and fizzy drinks.
- *Consuming foods that suit individuals with distributed meals:*

  Having multiple smaller meals instead of fewer larger ones may lower the likelihood of symptoms.

- *Prioritizing mental health:*

  Sufficient sleep and consistent physical activity enhance general wellness, making it easier to cope with Crohn's.

RECENT RESEARCH[10]:

- *Innovative Therapies:*

  Bibliometric analyses have shown that treatments like vedolizumab, ustekinumab, and Janus kinase (JAK) inhibitors are receiving more attention. These therapies are becoming more and more well-liked due to their unique processes and potential efficacy.

- *Cutting-edge Drug Trials:*

  Phase 3 studies of a monoclonal antibody developed by Teva Pharmaceuticals and Sanofi, were initiated after a Phase 2B research showed promising remission rates in patients with Crohn's disease and ulcerative colitis.

- *Non-Invasive Therapies:*

  As a non-invasive treatment for IBD, vagus nerve stimulation (VNS) is presently being researched. Preliminary experiments have demonstrated symptom relief and reduced inflammation, offering patients a potential adjunctive treatment.

CONCLUSION:

Crohn's disease is a long-lasting, relapsing, and incurable inflammatory bowel disorder with a complex origin that includes genetic, environmental, microbial, and immune factors. It is marked by chronic inflammation of the digestive system, frequently necessitating continuous treatment with medication, changes in lifestyle, and, on occasion, surgical intervention. Although treatments may lead to remission and assist in symptom management, the condition continues to progress, presenting risks such as bowel blockages, malnutrition, and a heightened likelihood of colorectal sarcomas. Timely diagnosis, attentive multidisciplinary management, and tailored treatment strategies are crucial for minimizing complications, enhancing quality of life, and maximizing long-term results. Continuous investigation into tailored and personalized treatments is progressing management approaches for impacted individual

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Chapter - 2

A Study of Gene Responsible for Causing of Color Blindness

Ashwin. K

ABSTRACT:

Color blindness is an eye disorder which cause a person, unable to distinguish between different colors. Comprehension of the natural science behind color vision has progressed significantly through the use of molecular genetics techniques. Despite their complication, the biological mechanisms behind color vision are easier to study than those of numerous other neural systems. This is partial due to the abundance of genetic differences influencing color perception within and between species, and because aspects of the color vision system are amenable to genetic modification. Mutation and alterations in the genetic factor that code for long, medium, and short wavelength-sensitive cone pigments are associated with color vision deficits with identified alterations impacting cone type quantity, pigment absorption spectra, cone functionality and viability, and the arrangement of the cone mosaic.

Keywords: Colour blindness; gene , mutation, symptoms, diagnostics, Treatment

Introduction:

Color blindness (CB), refers to the inability or reduced capacity to distinguish color variations in standard lighting conditions. CB may be categorized as either inherited or acquired. The occurrence of inherited color blindness is around 8% in males and 0.4% in females, caused by changes or lack in the absorption band of photopigments[1]. The incidence of color blindness differs between various ethnic groups around the globe. A latest study conducted in Eastern India has indicated that 8.73% of males and 1.69% of females are color blind[2].

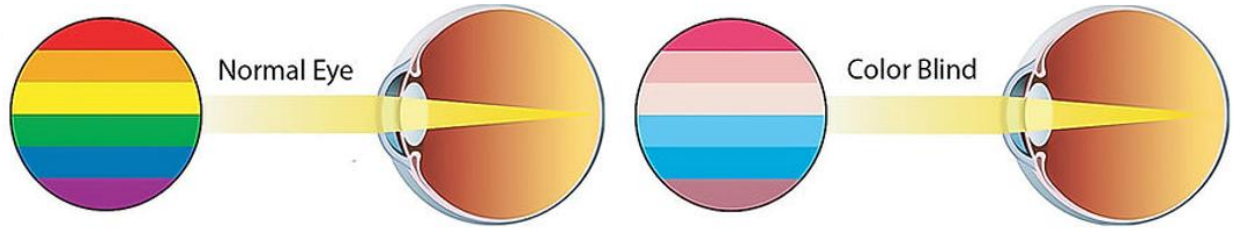

Figure 2.1: Understanding of color between normal eye and color-blind eye.

Genetics[3]*:* Color blindness is frequently passed down from one or both parents. This occurs because the genes linked with color blindness present in X chromosome

Condition for a male to be colour blind: Males possess a single X chromosome while females have a pair. A woman who possesses the gene has a son with a 50 percent probability of being color blind. The mother does not have color blindness since the gene is recessive. This indicates that her dominant normal gene overrides its influence.

Condition for a female to be colour blind: A daughter typically will not be color blind but will be a carrier unless her mother is a carrier and her father has color blindness.

GENE CAUSING COLOUR BLINDNESS[3]:

OPN1LW and OPN1MW genes:

The genes encode for the opsins associated with long wavelength (red) and medium wavelength (green), respectively. Alterations in these genes can result in red-green color blindness. This gene gives directions for producing a protein known as long wave sensitive opsin, which reacts to light in the yellow or orange section of the visible spectrum. This protein is located within the membrane of the cone cells found in the retina and assists in sensing red light. This gene is situated on the X chromosome and belongs to a tandem array containing one or more genes

OPN1SW gene:

This gene encodes for blue opsin, which has a short wavelength. Alterations in this gene can result in blue yellow color vision deficiency. This type of colour blindness can prevent a person from seeing and distinguishing between blue and yellow hues. This gene mutation causes color blindness, allowing the individual to perceive the world in hues of blue and red, while preventing them from distinguishing yellow from other colors by causing their eyes to interpret yellow as either blue or a blue shade.

CAUSES OF MUTATION IF THE GENE[4]:

1. Recombination events: During meiosis, misalignment of the OPN1LW and OPN1MW genes may lead to the formation of hybrid genes or additional copies of the genes. This additional copy should be removed or the alignment needs adjustment to resolve the issue.

2. Point mutations: Alterations in the DNA sequences of genes can influence the activity of the opsin protein. The nucleotide base is altered, resulting in a change to the amino acid required for the gene's and proteins normal function.

3. Copy number variations: Variations in the quantity of gene copies can also affect color vision. The quantity of a gene's copies is crucial because even a slight alteration in number can result in the gene synthesizing a completely different protein, potentially causing gene dysfunction.

4. Chemical mutagens: Certain substances such as 5-bromouracil can directly harm DNA sequences and can disrupt the replication process.

5. Physical mutagens: Types of radiation or physical stress such as Ultra–Violet (UV) radiation or Ionizing radiation (X-Ray or Gamma Rays) cause double strand breaks, deletions, and chromosomal rearrangements, potentially distorting DNA structure.

Figure 2.2: Color variations

EFFECTS OF MUTATIONS[4]:

Red green colour blindness: One of the main effects of the mutation is that the individual is unable to differentiate between the colors red and green. Finding careers that involve color difference and those that depend on color, such as fashion design and the textile sector, is another difficulty for those with color blindness. These mutations affect the traits and daily lives of typical people, making them pickier about what they wear and what trendy things to wear.

SYMPTOMS[5]:

The main sign is the absence of full color vision. But it's more complex than simply not being able to see specific colors with most kinds. A person with color vision impairment may have trouble with:

- Differentiating between particular hues
- Seeing the full range of color shining or richness
- noticing variations or tones of a single color

DIAGNOSIS[6,7]:

1. Ishihara Test: One often used method for detecting color blindness is the Ishihara Color Test. This evaluation, which has the name of its creator, Dr. Shinobu Ishihara, consists of a set of plates with colored dots arranged in particular patterns. Persons with standard color vision can easily identify the figures or shapes in the patterns, but people with color vision impairments may have trouble. The Ishihara test is a common tool used by ophthalmologists and optometrists in eye exams and is helpful in detecting various types of color vision deficits.

2. Basic Color Test: This test assesses a person's ability to distinguish between different hues. Typically, it involves identifying and matching various hues, shapes, or numbers present in colorful patterns. This assessment aids in evaluating the level of color perception and any shortcomings in distinguishing particular hues. It serves as a basic evaluation for comprehending an individual's Colour vision abilities and frequently forms a component of thorough eye assessments.

3. Sanjeevan Test: The Sanjeevan Colour Test includes 24 Color shades, with each shade signifying a unique Color for the patient to recognize precisely. The person is directed to align the given Colour, aiding in evaluating Colour recognition skills.

4. Enridge Green Lantern Test: The examination shows 9 sets of lights arranged vertically, each showing combinations of red, green, or yellow. The assignment is to recognize the colors present both above and below, with some potentially being identical. The Colors are seen for just two seconds. If the person has trouble recognizing the Colors, just press the next button.

Examples: 1. Up – red, down – green 2. Up – yellow, down – red.

TREATMENTS[8]:

Currently there are **no medications** that can cure colour blindness. However, researchers are exploring potential treatments. Potential Treatments that can help to battle colour blindness are:

1. Stem cell therapy: Stem cells are genetically identical cells that have the capacity to developing into differentiate into many specialized cell types, which play a role in repairing or replacing damaged tissues or cells due to a disease or disorder. The main objective of stem cell therapy is to utilize the healing and restoring abilities of stem cells to address or control various medical issues.

2.Gene therapy : This biotechnological method involves the introduction of normal genes into a person's cells to address genetic disorders. Healthy genes are introduced into the patient's cells typically through vectors such as viruses to replace the absent or defective gene function. This approach has demonstrated potential in addressing multiple conditions such as genetic diseases, certain sarcomas s, and viral infections. There are obstacles such as immune reactions to the inserted gene and making sure the gene therapy is directed to the particular cells impacted by the illness.

3. Assistive Technologies Colour Correcting Glasses or Lens: Both devices are designed to improve colour perception and function in quite similar manners. The lens and glass utilize specialized notch filters to obstruct the particular wavelength of light where the faulty wavelength of color coincides

RECENT RESEARCH[9, 10]:

Recent studies on color blindness focus on advancements in possible therapies, especially via gene therapy, and exploring how color vision deficiency affects quality of life.

1. Effect on quality of life: Studies investigate how color vision impairment influences daily living, emotions, and employment. Instruments such as the CVDQoL questionnaire are utilized to evaluate the effect on quality of life.

2. Filters and visual aids: Research examines the impact of specific filters in eyewear or tinted contact lenses on improving color perception and how this may change with color blindness.

3. Comprehending the brain's functions: Studies investigate how the brain interprets color data and how it may adjust in cases of color blindness

4. Retinal implants and surgery: To replace injured cone cells, researchers are creating retinal implants that convert light into electrical signals that the brain can understand as color information. Presently under research, these implants have the potential to transform the lives of people who are affected by color blindness.

CONCLUSION:

A color blindness is mainly brought on by genetic changes in the cone opsin gene. Red-green deficits are caused by the most common variations, which involve the OPN1LW and OPN1MW opsins. Uncommon tritan deficiencies are caused by mutations in the blue cone opsin gene. Cone sensitivity is altered by these abnormalities, impairing normal color perception. Color vision tests are essential for the first diagnosis. Advanced methods, such genetic testing, provide precise identification of the causative mutations. Supportive devices like specialized lenses and applications can help, even though there isn't currently a permanent cure. Research into gene therapy offers hope for future medical interventions. Although color blindness usually has no effect on general health, it can affect some jobs and daily activities.

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Chapter - 3

Polycystic Ovary Syndrome (PCOS): A Comprehensive Review

Durga. H

ABSTRACT:

Polycystic Ovary Syndrome (PCOS) is a predominant hormonal complication in women, but it remains one of the most frequently undiagnosed illnesses. Factors such as lifestyle or diet, environmental toxins, genetics, gut microbiome imbalance, neuroendocrine changes, and obesity contribute to the risk of PCOS in females. These elements could lead to a rise in metabolic syndrome by inducing hyperinsulinemia, oxidative stress, hyperandrogenism, disrupted folliculogenesis, and abnormal menstrual cycles. Sadly, though, many women with PCOS remain unaware and uninformed about their condition. PCOS affects 8–13% of women in their generative years, influences biopsychosocial factors, and imposes a considerable economic burden on health related to reproductive, metabolic, and psychological complications. Being a complex and multifaceted condition, it affects women's health and well-being in numerous ways. Thus, it is essential to raise awareness and highlight preventive measures

Keywords: PCOS, Causes, Symptoms, Treatment

INTRODUCTION:

Polycystic Ovarian Syndrome (PCOS), is a hormonal disorder that results in engorged ovaries featuring small cysts around the periphery. Polycystic Ovary Syndrome (PCOS) is a prevalent endocrine condition impacting those with ovaries, frequently resulting in hormonal imbalances and metabolic issues[1,2]. It is marked by signs like abnormal menstrual patterns, high androgen levels, and the existence of numerous small cysts in the ovaries. It is believed that both hereditary and environmental factors play a significant role in PCOS, even though the exact etiology is still unknown[3].

PCOS is used to describe the progress of several small cysts in the ovaries[4]. The World Health Organization (WHO) states that 8–13% of women in their reproductive years suffer from PCOS, a chronic multifactorial condition. One in ten women of reproductive age have PCOS, making it one of the leading causes of infertility globally[5]. The good news is that up to 80% of women with PCOS can become pregnant with the correct medical attention and lifestyle changes[6]. According to diagnostic criteria, the prevalence rate among teenagers was 22.6%, and

the rate of androgen excess was 9.8%. In India, the estimated prevalence is 3.7%. Higher than usual quantities of androgens, or male hormones, are produced by the ovaries in PCOS, which can interfere with ovulation and upset the menstrual cycle[7].

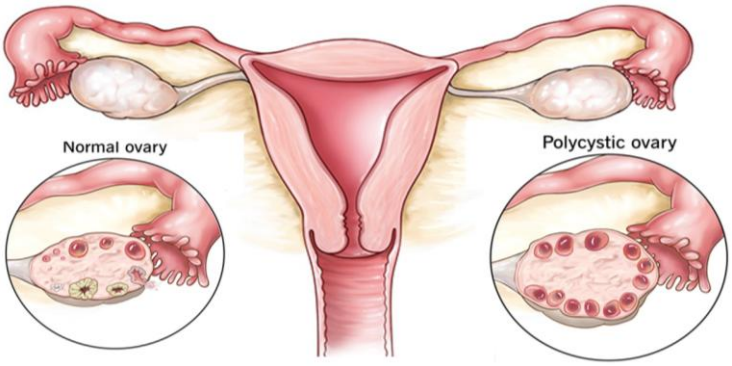

Figure 3.1: Showing the difference between Normal and PCOS ovary

SYMPTOMS OF PCOS[8,9]:

Different people may display different illness signs. Some of these symptoms include:

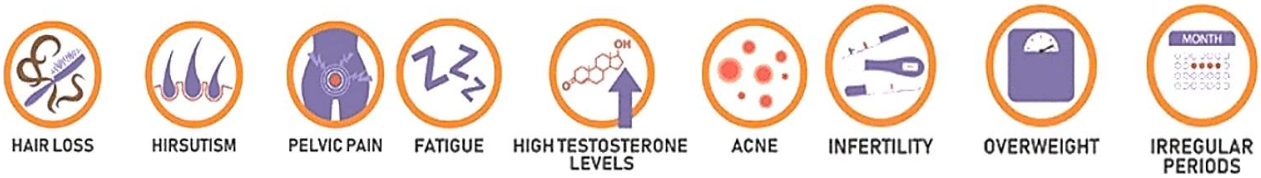

Figure 3.2: Symptoms of PCOS

CAUSES OF PCOS[10,11]:

Figure 3.3: Causes of PCOS

TREATMENT TOWARDS RECOVERY[12,13]:

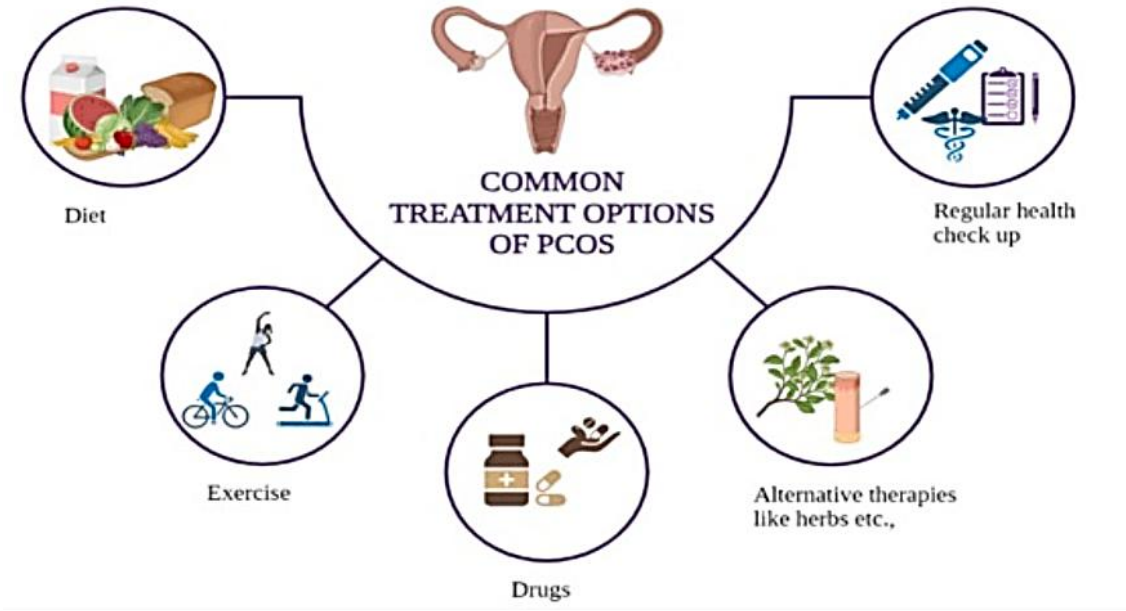

Figure 3.4. Treatment Towards Recovery

DRUGS[14]:

Table:3.1. Drug and their function for PCOS

Drug	Function
Metformin	Enhancing insulin sensitivity, controls blood glucose levels
Clomiphene Citrate and Letrozole	To stimulate ovulation in women facing infertility from PCOS.
Oral Contraceptives- Combination pills (ethinyl estradiol and progestin)	Managing menstrual cycles and lowering androgen levels
Anti-Androgen Drug- spironolactone	Reducing excessive hair growth and acne caused by elevated androgen levels.

DIET:

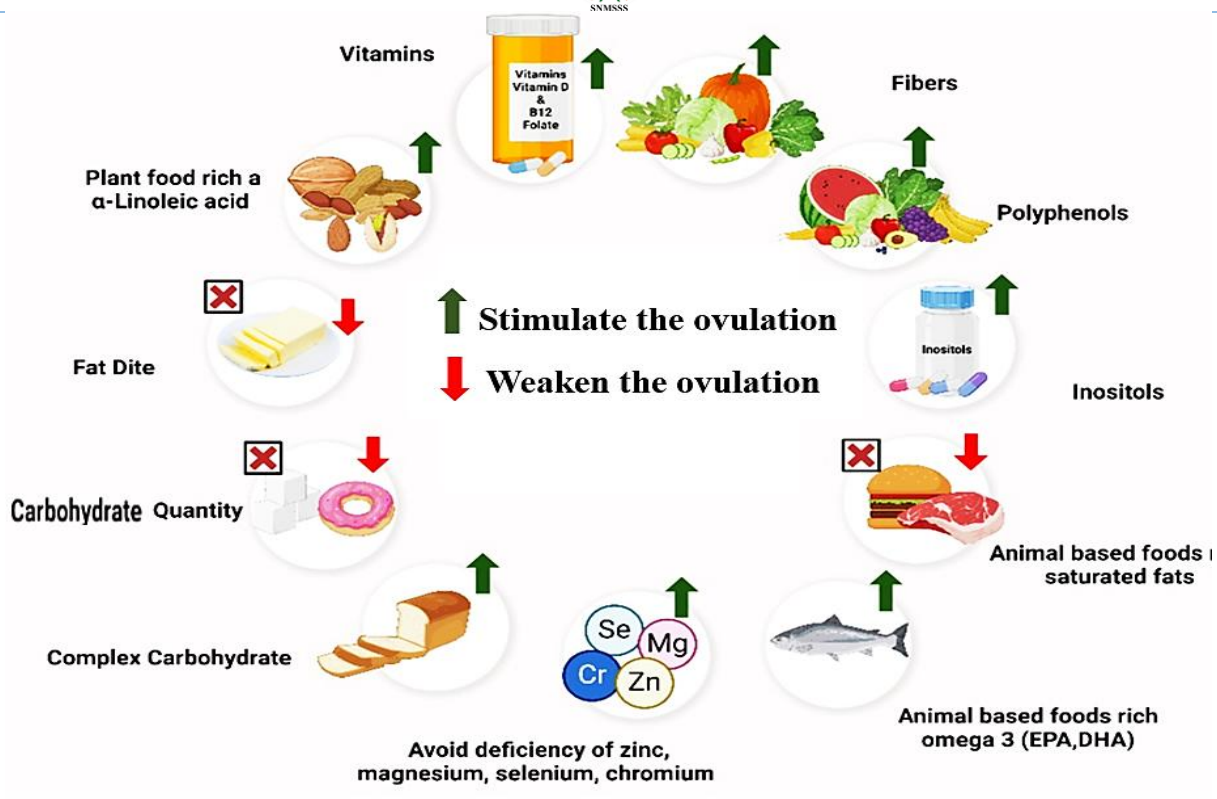

Figure 3.5. Diet PCOS

DIAGNOSIS OF PCOS:

Figure 3.6. DIAGNOSIS OF PCOS

CONCLUSION:

Polycystic ovary syndrome increases the threat of enduring health problems like type 2 diabetes, heart disease, and obesity in addition to causing irregular menstruation periods and infertility. Furthermore, the emotional repercussions, which range from anxiety to despair, highlight the need for all-encompassing care that takes into account mental and physical health. Understanding the relationship

between cortisol levels and chronic stress emphasizes the importance of lifestyle management, which includes stress-reduction techniques, regular exercise, and a balanced diet. To effectively manage PCOS and improve the quality of life for people affected, prompt diagnosis, education, and teamwork are crucial.

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Chapter - 4

A Review of the Literature on Pelvic Bone Sarcomas

Disha. A

ABSTRACT:

Primary bone sarcomas are uncommon malignant tumors of mesenchymal origin. The outlook has advanced over the years, but bone sarcomas continue to be dangerous tumors requiring a multidisciplinary strategy for diagnosis and treatment. Pelvic bone sarcomas pose a distinct challenge for orthopedic oncologists because there are no natural anatomical confines, essential neurovascular structures are located nearby, and pelvic reconstructions after tumor removal endure significant mechanical stress. Although radiotherapy plays a significant role in osteosarcoma, surgery continues to be the primary treatment option. Although external hemipelvectomy is still one option, the primary goal of surgery is to preserve the limb. Following the total removal of the tumor, the bone defect must be repaired. Options for repairing the flaw involve prosthetic or biological modernization. The approach to reconstruction relies on the cancer's position and the type of surgery needed for its extraction. This article aims to provide an overview of pelvic bone sarcomas and treatment alternatives

Keywords: Sarcomas, Types, Causes, Treatment, Advance Development.

INTRODUCTION:

Sarcomas of the pelvic bone are an uncommon and serious condition marked by malignant tumors located in the pelvic area's bones, such as the ilium, ischium, pubis, and sacrum. This type of sarcoma presents significant difficulties because of the intricate structure and role of the pelvic girdle, which acts as the weight-bearing foundation of the spine and accommodates essential organs like the bladder, rectum, and reproductive organs[1,2]. Approximately half of all bone tumors are non-cancerous. About 5–10% of osteosarcoma instances primarily arise in the pelvic bones. Research on survival rates for primary pelvic osteosarcoma indicates that the 5-year survival rate ranges from 15–30%, contrasting with the 60–70% rate noted for osteosarcoma overall[2,3].

Figure 4.1: Showing cancer cells in the plvic bone

Cancerous tumors that originate mainly from bone cells are uncommon. These tumors typically emerge in individuals between the ages of ten and twenty. They can originate from bone cells as well as from cartilage cells. Depending on its severity, the tumor may closely resemble the original tissue or be hardly recognizable as it[4]. Osteosarcoma is a cancerous tumor that typically occurs in males aged 10 to 25 and generally impacts the long bones. Ewing's sarcoma primarily impacts young people. In this instance, the femur, pelvis, fibula, tibia, and humerus are commonly affected. Primarily, individuals aged 40 to 60 years are impacted, with the pelvic region being the most frequent site[5].

However, most bone tumors do not arise from the bone itself; rather, they are metastases. These metastases primarily originate from the breast, kidneys, lungs, or prostate. The majority of bone metastases decompose the bone material and harm the bone structure. This makes the affected bones weak and can lead to breaks[6]. A few bone metastases lead to an increase in bone tissue density. Additionally, metastases can destroy bone in certain areas while increasing its density in other areas[7].

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TYPES OF PELVIC BONE SARCOMAS[3,8]:

1. Primary Pelvic Bone Sarcomas: These sarcomas arise directly from the pelvic bones.

a) Osteosarcoma: The most prevalent primary bone sarcoma, originates from cells that form bone, frequently impacts adolescents and young adults, aggressive and grows quickly.

b) Chondrosarcoma: Develops from cells that produce cartilage, More prevalent in individuals over 40, Gradually increases in size but may be unresponsive to chemotherapy and radiation.

c) Ewing's Sarcoma: Generally impacts children and young adults, Can develop in the pelvis or long bones, Very aggressive but reacts positively to chemotherapy and radiation.

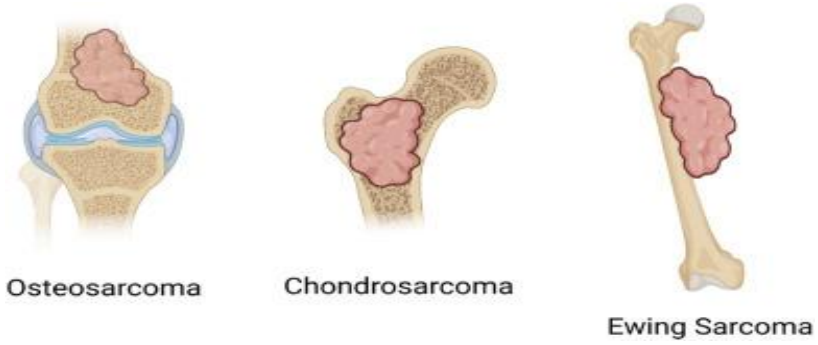


Figure 4.2: Represent types of pelvic bone sarcomas

2. Secondary (Metastatic) Pelvic Bone Sarcomas: These begin in different parts of the body but move (metastasize) to the pelvic bones.

Typical origins include: Prostate sarcomas (in males), Breast sarcomas (in females), Lung sarcomas, Kidney sarcomas, Thyroid sarcomas. The pelvis frequently serves as a metastasis site because of its abundant blood supply and bone marrow presence.

3. Hematologic Bone Sarcomas (impacting the pelvis): These sarcomas target the bone marrow located in the pelvic bones.

a) Multiple Myeloma: Tumors of plasma cells in the bone marrow, Can weaken pelvic bones and lead to fractures or discomfort.

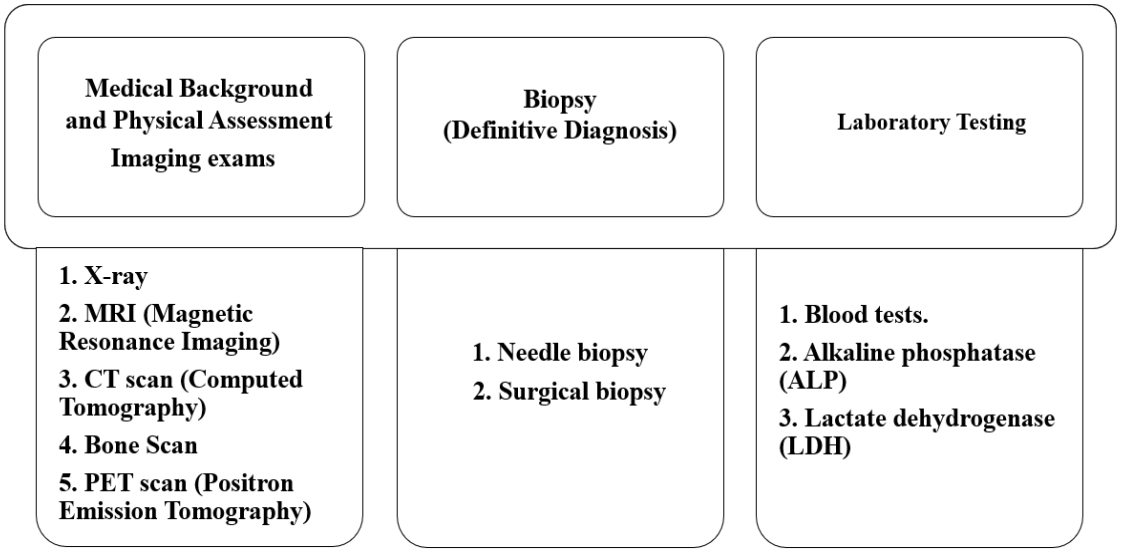
b) Bone lymphoma: Rare kind of lymphoma that can begin in the pelvic bones or spread there.

CAUSES OF PELVIC BONE SARCOMAS[6-8]:

Table 4.1 Causes of Pelvic Bone Sarcomas

Genetic Factors	Rare genetic disorders that are inherited and increase the risk of bone cancer
Past Radiation Therapy	Experience bone cancer in areas exposed to radiation, such as the pelvis
Bone Disorders	Cause malignancies to develop in the bone
Swift Bone Development	Adolescents during times of rapid growth are the most common variant.
Chemical Exposure	Alkylating drugs are a risk factor for malignancies that have already occurred.

DIAGNOSIS OF PELVIC BONE SARCOMAS[9]:



Flow Chart: 4.1. Diagnosis Of Pelvic Bone Sarcomas

Staging assessment[9]:

- Upon confirmation, imaging is employed to assess the stage (degree of spread).
- Staging assists in directing treatment choices.

SYMPTOMS OF PELVIC BONE SARCOMAS[10]:

Table 4.2. Symptoms of Pelvic Bone Sarcomas

AREA	SYMPTOMS	RESULT
Chronic Pelvic or Hip Discomfort	Constant, throbbing pain in the pelvis, hips, or lower back	Intensifies at night or while at rest.
lump or swelling	Detectable bump might appear in the hip, lower back, groin, or buttocks, and the swelling	Not cause pain at first.
Limping / Walking Difficulties	may impact leg muscles and joints	resulting in a changed gait or limping.
Limited Movement, Numbness or Tingling	Rigidity or decreased mobility in the hip or leg	Discomfort.
Bowel / Bladder Issues	Constipation, Urinary retention or leakage	tumor development can influence pelvic organs
Pathologic Fractures	Pain even from a slight injury.	Bone Fracture in any parts of Body

TREATMENT OF PELVIC BONE SARCOMAS[9,10]:

1. Surgery: To prevent recurrence, remove the tumor with distinct margins. often requires complex pelvic reconstruction because of the structure of the pelvis, which may include bone transplants or metal implants.

Categories of surgical procedures:

- Limb-preserving surgery: Eliminates tumor while maintaining limb functionality.
- Pelvic resection: Could entail excision of a section of the pelvic bone.

2. Chemotherapy: Uses drugs to eliminate sarcoma cells or inhibit their proliferation.

- Commonly used for Osteosarcoma: Methotrexate (high-dose), Doxorubicin (Adriamycin), Cisplatin, Ifosfamide
- Commonly used for Ewing's Sarcoma: Vincristin, Doxorubicin, Cyclophosphamide, Etoposide (sometimes used), Pazopanib (a targeted therapy medication for soft tissue sarcomas, at times used off-label), Denosumab (applied in certain instances to prevent bone damage)

3. Radiation Therapy: Employs high-energy X-rays to terminate sarcoma cells. Commonly employed for:

- Tumors that cannot be surgically excised Ewing's sarcoma
- When surgery is not fully accomplished or cannot be performed, it may be paired with chemotherapy.

4. Targeted Therapy: Recent treatments that focus on particular molecules involved in the growth of sarcomas, utilized in certain situations according to tumor genetics.

5. Supportive Care: Managing pain, engaging in physical therapy, and rehabilitation to enhance life quality. Emotional assistance and diet management.

6. Clinical Trials: Patients might have opportunities to engage in studies evaluating new medications or treatment combinations.

The treatment of pelvic bone sarcomas is intricate and necessitates a team of specialists: oncologists, orthopedic surgeons, radiologists, pathologists, and rehabilitation experts

ADVANCE DEVELOPMENT[11,12]:

1. Improved Surgical and Imaging Methods:

Table 4.3. Improved Surgical and Imaging Methods

Techniques	**Effect**
Navigation and 3D Planning	Enhanced resection precision - safeguarding essential nerve and vascular elements.
Limb Salvage	Internal hemipelvectomy - Resulting in favorable functional outcomes.
Reconstruction Techniques	Relocating the femur to connect with the remaining ilium or sacrum.

2. Enhanced Proton Beam Therapy in Radiotherapy:

Advances in radiotherapy, particularly proton beam therapy, allow for the delivery of larger, more effective doses (>60Gy) to the tumor with less damage to nearby healthy tissues for pelvic sarcomas that cannot be surgically excised or are only partially removed.

3. Focused and Novel Treatments:

- Molecular Targeted Agents: New therapies are being investigated for advanced, metastatic, or unresectable conditions. Instances involve the application of ivosidenib for IDH1-mutated, locally advanced, or metastatic typical chondrosarcoma (CHONQUER study).
- Tyrosine Kinase Inhibitors (TKIs): Substances such as regorafenib and cabozantinib have demonstrated potential in postponing disease advancement in advanced/metastatic osteosarcoma.
- Immunotherapy: For the specific bone sarcomas, research on checkpoint inhibitors is still in its early phases.

4. Centralized Healthcare Services:

Table 4.3. Centralized Healthcare Services

Multidisciplinary Teams (MDT) oncologists in orthopedic, radiologists, pathologists	Enhancing survival rates in patients
Dedicated Centers (personalized, evidence-driven treatment)	Considering the intricate, low-volume characteristics of the tumors.

CONCLUSION:

Sarcomas in the pelvic bone are rare, because of the complex structure of the pelvic region, but potentially lethal conditions that pose significant challenges to diagnosis and treatment. To improve patient survival and quality of life, early detection through imaging and biopsy is essential, as is a team-based treatment plan that includes radiation therapy, chemotherapy, and surgery. Advancements like as genetic fingerprinting, precision medicines, and 3D-printed implants have revolutionized surgical reconstruction and personalized care, giving patients new hope. In spite of the potential risks and side effects linked to treatment, continuous research and clinical trials are improving results and broadening therapeutic options. In the end, increased awareness, timely diagnosis, and advanced medical treatments are vital for alleviating the challenges posed by pelvic bone sarcomas and promoting improved patient care

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Chapter - 5

Predicting the Mutation, Molecular and Cellular Level of the CFTR Gene Responsible for Cystic Fibrosis Using NCBI Database

Keerthana. R

ABSTRACT:

Cystic Fibrosis (CF) is a multi-organ disease mainly affecting the lungs, defined by airway blockage, infections, and inflammation. More than 75% of CF patients are identified by the age of 2, with a majority surviving past 18 years. The condition is associated with more than 2,000 mutations in the CFTR gene. Present therapies target CFTR modulators that boost CFTR protein activity and enhance lung condition. Nonetheless, these treatments encounter obstacles, such as limited effectiveness against various CFTR mutations, high expenses, and worries regarding long-term safety and efficacy. This review examines the genotypic manifestation of CFTR and assesses existing therapeutic approaches. More accurate and long-lasting therapies could reduce the incidence of CF, increase mutation coverage, and improve treatment efficacy with these cutting-edge methods.

Keywords: CFTR mutations, Screening, Therapies, Cystic fibrosis

INTRODUCTION:

Cystic Fibrosis is an autosomal recessive disorder mainly affect respiratory and digestive systems, which triggered by mutations in the Cystic Fibrosis Transmembrane Regulator (CFTR) gene. The disease affects 40,000 individuals in the US and 105,000 globally, showing a significant rise in both incidence and survival rates[1, 2]. In 1989, identification of the gene (CFTR) brought optimism to the research and medical fields, resulting in the revealing of more than 2000 distinct mutations. This can enhanced analysis and attention for CF patients and their folks and paved the way for developing remedy targeted at particular mutations[3].

CF harms various organs, resulting in advancing lung deterioration and death. Improvements in medical care and therapies have increased the average life expectancy of CF patients to the early 50's. Recent progress in drug therapies, especially CFTR modulators, enables the aiming of the genomic basis of CF mutations, which may restore mutant CFTR role and enhance the value of life for CF patients[4].

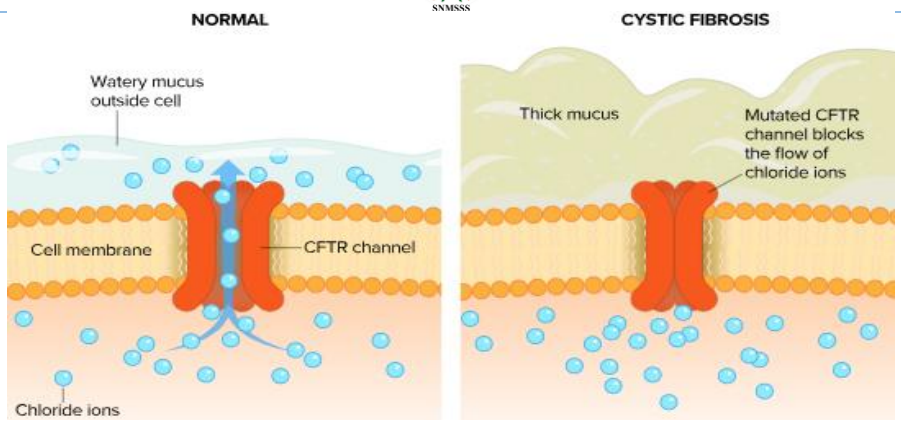

Figure 5.1. Showing different between the function of normal and mutated CFTR

CAUSES OF CYSTIC FIBROSIS[5]:

Mutation in the CFTR Gene: The primary cause of CF is a mutation in the CFTR gene. This gene responsible for guidance to produce a unique protein that controls the flow of salt and H_2O in and out of cells. This function is crucial for maintaining mucus in the body as thin and slippery. The mutation affects the CFTR gene, it results in the formation of a malfunctioning CFTR protein or no protein whatsoever. Consequently, the equilibrium of salt and water within the body cells is disrupted. This results in mucus becoming thick and adhesive, accumulating in the lungs, digestive tract and other organs, resulting in the signs of CF.

SYMPTOMS OF CYSTIC FIBROSIS[6]:

Table 5.1 Symptoms of Cystic Fibrosis

Respiratory systems	Digestive systems
• Recurrent or chronic lung infections • Cough (associated with gagging, vomiting and disturbed sleep) • Thick sputum (mucus) production • Whistle-like sound production on breathing (Wheezing) • Extreme exhaustion from exercise • Stuffy nose • Recurrent sinusitis	• Feeling of bloating or abdominal tightening • Failure to pass meconium (in a newborn) • Episodes of constipation • Crampy abdominal pain • Change in stooling pattern • Decreased appetite • Vomiting • Foul-smelling, greasy stools • Poor weight gain and growth

CFTR GENE Predicted from NCBI Database:

Mutation in the CFTR Gene:

LOCUS KU325498 568 bp DNA linear PRI 05-JUL-2016

DEFINITION Homo sapiens CFTR (CFTR) gene, partial cds.

CHROMOSOME : 7

ACCESSION : KU325498

VERSION : KU325498.1 .

SOURCE : Homo sapiens(human) ORGANISM Homo sapiens

Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria;

Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo.

FEATURES Location/Qualifiers

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FASTA Sequence of CFTR :

>KU325498.1 Homo sapiens CFTR (CFTR) gene, partial cds

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GCAAAAGGAAGGGGTGGTGTGCGGAGTAGGGGTGGGTGGGGGGAATTGGAAGCAA
ATGACATCACAGCAGGTCAGAGAAAAAGGGTTGAGCGGCAGGCACCCAGAGTAGT
AGGTCTTTGGCATTAGGAGCTTGAGCCCAGACGGCCCTAGCAGGGACCCCAGCGCC
CGAGAGACCATGCAGAGGTCGCCTCTGGAAAAGGCCAGCGTTGTCTCCAAACTTTTT
TTCAGGGGAGAAGGTGGCCAACCGAGCTTCGGAAAGACACGTGCCCACGAAAGAG
GAGGGCGTGTGTATGGGTTGGGTTTGGGGTAAAGGAATAAGCAGTTTTTAAAAAGA
TGCGCTATCATTCATTGTTTTGAAAGAAAATGTGGGTATTGTAGAATAAAACAGAAA
GCATTAAGAAGAGATGGAAGAATGAACTGAAGCTGATTGAATAGAGAGCCACTTCC
TACTTGCAATAGAGAGCCACCATCCTCCTTGCTCCACATGTGGCCCCTTTTCACTTG
TCATCCTG

MOLECULAR AND CELLULAR-LEVEL FACTORS OF CFTR[7]:

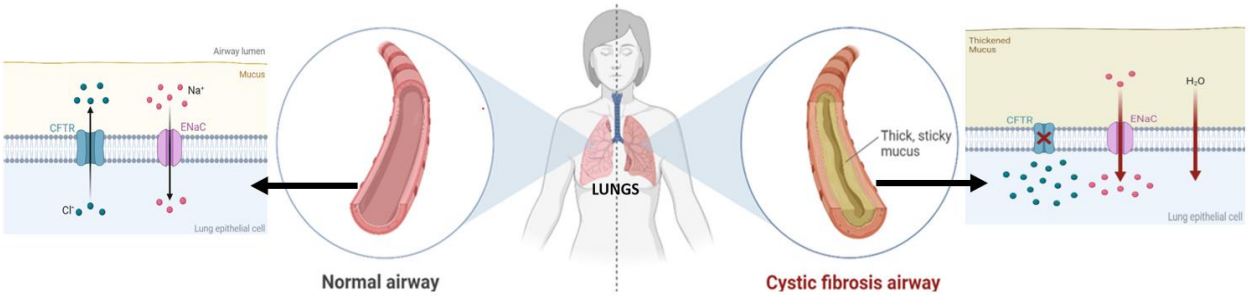

Figure 5.2. Representing different Molecular and cellular function of normal and mutated CFTR

The CFTR protein transports Cl- from a cell's interior to its outside. Its purpose is to maintain a healthy coating of mucus between the body and the external environment by balancing salt and water. Mucus in the lungs collects material, including dust, bacteria, and viruses. To remove this detritus from the body, cilia oscillate back and forth. The mucus layer in the lungs becomes thick and sticky if the CFTR protein is impacted, which causes an imbalance between salt and water.

DIAGNOSIS AND SCREENING OF CYSTIC FIBROSIS[8]:

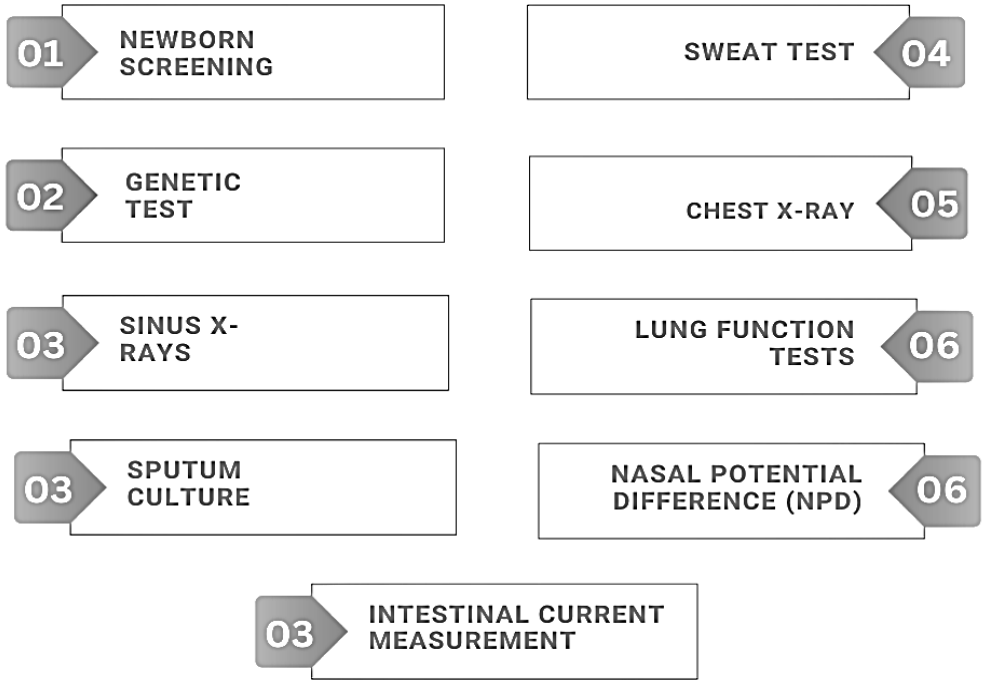

Figure 5.3. Diagnosis and Screening of Cystic Fibrosis

CFTR Modulator Therapy[9]:

Table 5.2 CFTR Modulator Therapy

Therapy	Drug Name	Uses
Mucus Thinning and Clearance	Dornase alfa	Enzyme that breaks down DNA in mucus to reduce its thickness
Bronchodilator	Albuterol - Salbutamol	Beta-2 agonist that eases breathing
Antibiotic	Tobramycin	Inhaled antibiotic targeting *Pseudomonas aeruginosa.*
	Aztreonam lysine	Used via inhalation for chronic infections
Anti-Inflammatory	Azithromycin,	Used for its long-term anti-inflammatory effects

Additional treatment:

Physical and Airway Clearance Techniques: These non-pharmacological methods include oscillating vests, breathing apparatuses, and chest physical therapy to physically remove mucus from the lungs.

Lung Transplantation: Lung transplantation is required when a patient has end-stage cystic fibrosis and has significantly diminished lung function. After this intricate surgical procedure, patients must take medication for the rest of their lives. Following a lung transplant, medications such as tacrolimus, prednisone, and mofetil mycophenolate are used to reduce immunological rejection.

RESEARCH AND FUTURE DIRECTIONS[5,10]:

1. Gene Therapy and Gene Editing: - Researchers are aiming to provide a healthy version of the CFTR gene to the body through viral or non-viral delivery systems. The objective is to fix or substitute the defective gene so that cells can generate active CFTR protein. A promising approach is CRISPR-Cas9, a gene-editing mechanism that could accurately fix the CFTR mutation within the DNA.

2. *mRNA Therapy:* - mRNA treatment for CF includes providing a version of messenger RNA that encodes the correct CFTR protein. The cell utilizes this mRNA to create a functional variant of the protein for a limited time. In contrast to gene therapy, mRNA therapy does not modify the DNA but provides the body with the necessary instructions to generate a healthy CFTR protein.

3.New and Improved CFTR Modulators: - Although medications such as Ivacaftor and the triple therapy (Elexacaftor/Tezacaftor/Ivacaftor) have enhanced patient outcomes, scientists are creating next-generation CFTR modulators that aim to target rarer mutations and increase efficacy with reduced side effects. These new compounds are intended to be more efficient and beneficial to a greater range of patients.

4. Stem Cell Research: - Stem cells provide promise for restoring damaged lung tissue and potentially fixing the CFTR gene in a patient's own stem cells prior to reinserting them into the lungs. This may result in tailored and enduring therapies that extend beyond merely managing symptoms.

5. Targeting Inflammation and Infections: Novel treatments and drugs with anti-inflammatory qualities are being created to treat persistent bacterial infections. These include novel antibiotics, anti-biofilm agents, bacteriophage medications that decrease the bacterial response and viruses that kill bacteria.

6. Artificial Intelligence and Personalized Medicine: Researchers can now predict drug reactions, disease development, and the best treatment combinations for each patient by using AI and machine learning. Every patient will receive treatment according to their unique genetic mutation and health status thanks to personalized medicine.

CONCLUSION:

Despite significant advancements in the use of CFTR genes to treat cystic fibrosis, significant challenges still exist. Many mutations are not addressed by current medications, leaving a considerable portion of the CF patients without effective cure selections. Gene therapy encounters real challenges such as expensive treatments, effectiveness concerns, and potential off-target properties. Restricted enduring safety and effectiveness records, challenges in delivery and ethical issues hinder its use. To address these challenges, future gene modulators should focus on non-responsive mutations, stem cell therapies, and single-dose treatment strategies such as genetic alterations.

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Chapter - 6

Leukemia – Review on Recent Developments in Treatment and Management

Kanika Srinivasan

ABSTRACT:

Leukemia is a type of blood cancer marked by the swift proliferation of a typical blood cell. The unregulated expansion occurs in bone marrow, where the majority of the body's blood cells are produced. Leukemia cells are typically underdeveloped (not fully mature) white blood cells. Leukemia is derived from the Greek terms"white" (leukos) and "blood" (haima). In contrast to other cancers, leukemia typically doesn't create a mass (tumor) that appears in imaging examinations, like X-rays or CT scans. Numerous varieties of leukemia exist. Some are more prevalent in children, whereas others are more in adults. The approach to treatment varies based on the specific type of leukemia and additional coexisting factors. This article seeks to summarize advancements in the management and treatment of leukemia.

Keywords: Leukemia, Causes, Management, Treatment.

INTRODUCTION:

Blood cancer includes a vast array of various malignancies. This category comprises cancers that affect the bone marrow, blood, and lymphatic system, which incorporates lymph nodes, lymphatic vessels, lymphoid tissue, thymus, tonsils and spleen in the digestive tract. Leukaemia and myeloma, originating in the bone marrow, along with lymphoma, originates in the lymphatic system, are the most prevalent forms of blood cancer[1]. Blood cancers impact the generation and functioning of blood cells. The majority of these cancers originate in the bone marrow, the site of blood production. Stem cells found in bone marrow grow and differentiate into three kinds of blood cells: RBC, WBC, or platelets[2]. In many blood cancers, the typical development of blood cells is disrupted by the unchecked proliferation of an atypical kind of blood cell. These irregular cancerous cells, hinder the blood from executing numerous routine functions, such as combating infections or stopping severe bleeding[3].

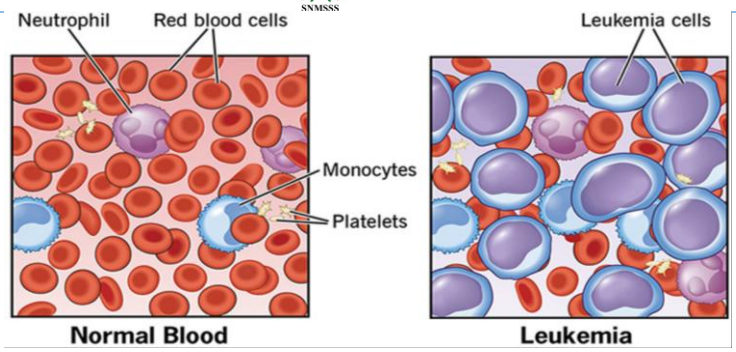

Figure 6.1: Showing the difference between Namal and Leukemia blood cell

CAUSES[4]: Leukemia initiates in the bone marrow, the soft, spongy tissue found in the internal cavity of skeletons produces blood cells. Blood cells include Platelets (Cells that assist in blood clotting), White blood cells (Cells that combat infection), Red blood cells (Cells that transport oxygen and essential substances to all tissues and organs throughout the body). These blood cells originate from hematopoietic (hemo = blood, poiesis = create) stem cells. Prior to the complete formation of blood cells, they pass through multiple developmental stages. The stem cells differentiate into myeloid cells or lymphoid cells. Leukemia is occasionally detected during blood tests conducted for another health issue. Researchers do not fully comprehend the precise origin of leukemia. It appears to arise from a mix of genetic and environmental influences[5].

Genes[5]:

Genetic conditions: Issues in an individual's genes can result in leukemia. For instance, individuals with specific genetic disorders such as Down syndrome are at an increased risk of developing leukemia. If an individual is a smoker or has been near smokers, they are at an increased risk of developing a form of leukemia.

Leukemia in family history: Certain forms of leukemia can be hereditary, though this is rare. Having a family member with leukemia does not mean that others will also develop it. If a family member has a genetic leukemia condition, the doctor can be notified. A doctor may recommend a genetic test to assess the risk.

Exposure to industrial chemicals: Benzene and formaldehyde are recognized as carcinogenic substances present in construction materials and domestic cleaners. Benzene is utilized in the production of plastics, rubber, dyes, pesticides, pharmaceuticals, and detergents. Formaldehyde is present in construction materials and home items like soaps, shampoos, and cleaning agents

SYMPTOMS[6]**:** Fever or shivering, Ongoing tiredness, Lethargy, Frequent or intense infections, Unintentional weight loss, Enlarged liver or spleen, Small red dots on the skin, Swollen lymph glands, Night sweats, Frequent nosebleeds, Easy bruising or bleeding, Bone discomfort or sensitivity

DIAGNOSIS[6]**:** Bone Marrow Biopsy, Positron Emission Tomography (PET) scan, Magnetic Resonance Imaging (MRI) scan, Computed Tomography (CT) scan, Complete blood count (CBC), Blood chemistry test, Blood cell examination.

TREATMENT[6-8]**:**

Leukemia is treatable with good success rate, and in certain instances, it may be completely cured. The chance of a cure of leukemia relies on numerous factors such as types, patient's age, general health, and the disease's response to treatment. Common treatments include:

I. Therapies:

1. Chemotherapy: Chemotherapy serves as a key treatment for blood cancers, destroying cancer cells to either hinder the progression of the disease or eradicate the cancer. Health care practitioners utilise various medication types for distinct blood cancers. Chemotherapy serves as a main treatment for blood cancer, destroying cancer cells to either hinder the disease's advancement or eradicate the cancer. Health care professionals utilize various medications for various blood cancers.

2. Radiation Therapy: A healthcare professional might utilise radiation to address leukaemia, lymphoma, or myeloma. Radiation focuses on abnormal cells, harming their DNA to prevent replication. Frequently, radiation therapy is combined with other therapies. They might utilise radiation to alleviate certain symptoms.

3. Immunotherapy: This approach utilises the patient's immune system to combat cancer. Immunotherapy can aid the patient's body in generating additional immune cells or assist the current immune cells in recognising and eliminating cancer cells.

4. Targeted Therapy: This cancer treatment focuses on genetic alterations or mutations that convert healthy cells into abnormal ones. Providers transform T-cell lymphocytes, a category of white blood cells, into more potent ones.

5. Induction therapy: Primary objective is to eradicate as many leukemia cells as feasible so that the blood cells and bone marrow cells can accomplish remission which indicates that blood cell counts return to normal, absents of leukemia cells in the bloodstream. The initial treatment phase, known as induction therapy, typically lasts 4 to 6 weeks

6. Maintenance therapy: Aim to eliminate any leukemia cells that might have persisted after the initial 2 treatment stages and avert the recurrence of symptoms (relapse). The duration of the treatment is approximately two years.

II. Transplantation:

7. Autologous stem cell transplant: Healthcare professionals can gather and preserve bone marrow stem cells prior to giving high levels of chemotherapy. After chemotherapy is completed, they will reintroduce the safeguarded stem cells. In this manner, individuals with autologous stem cell transplants can steer clear of chemotherapy adverse effects.

8. Allogeneic Stem Cell Transplant: Occasionally, impaired bone marrow must be substituted with healthy bone marrow. Medical professionals locate an appropriate bone marrow donor and utilize the donor's cells to replace the damaged tissue. This method is effective yet carries significant risks.

9. Stem Cell Transplantation: A stem cell transplantation introduces healthy stem cells (blood-forming) into the body. Stem cells can be sourced from the umbilical cord blood, peripheral blood, and bone marrow.

MANAGEMENT STRATEGIES[9]:

- **Minimal Residual Disease (MRD):** Evaluating MRD is crucial for determining treatment effectiveness and guiding prognosis.
- **Precision Medicine:** Therapy is progressively customized according to molecular profiling.
- **Reduced Intensity:** There is a shift towards lower-intensity, focused methods for unfit patients to lessen toxicity while preserving effectiveness, combining immune cells and sarcoma cells so that the immune cells can eliminate the sarcomas.

ONGOING STUDIES AND TESTS[10]:

- The Pediatric Preclinical *In Vivo* Testing Consortium (PIVOT) utilises mice to evaluate new treatments for childhood sarcomas, such as leukemia.

- To determine the best treatments for children and teenagers with acute lymphoblastic leukemia, the Children's Oncology Group carries out research.

FUTURE DIRECTIONS[10]:

- Combination treatments: combining chemotherapy with specific drugs.
- *Nanomedicine:* Innovative drug delivery systems are being created to enhance pharmacokinetics and minimise off-target toxicity in Leukemia.

CONCLUSION:

Research on leukemia has advanced remarkably in recent years, resulting in better diagnosis, treatment, and outlook. The landscape of treatment has evolved due to advancements in immunotherapy, targeted medicines, and genetic testing. However, challenges still exist, such as the need for improved treatments for particular leukemia types and improved access to healthcare for underserved populations. Ongoing research and teamwork are crucial for enhancing results for leukemia patients.

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Chapter - 7

A Review Rare Vasculopathy Disease – Moyamoya Disease

Monika. A

ABSTRACT:

Moyamoya disease is an unusual vascular condition affecting the blood vessels in the central nervous system, primarily the internal carotid arteries. It is a persistent obstructive cerebrovascular condition that is non-atherosclerotic and non-inflammatory. It is marked by hyperplasia of the endothelium and fibrosis in the intracranial segment of the carotid artery and its nearby branches, resulting in gradual stenosis and blockage, frequently presenting scientifically as hemorrhagic or ischemic stroke with elevated ill health and death rates. In cerebral angiography, the development of collateral vessels are more evident with advancements in contemporary imaging methods. When a disease is present, it is referred to as moyamoya syndrome. Current treatment options are restricted, but surgical revascularization might avert ischemic incidents and maintain quality of life. In this review, we outline the stages, diagnosis, and treatment of moyamoya disease

Keywords: Moyamoya disease, Stages, Management and Treatment.

INTRODUCTION:

Moyamoya disease (MMD) is a rare intracranial vascular disorder characterized by steadily narrowing of the blood vessels in the brain's base, leading to transient ischemic attacks or strokes[1]. The term moyamoya describes the puff of smoke appearance of small collateral vessels visible in the basal ganglia and thalamus, which develop in reaction to the progressive narrowing and blockage of the supraclinoid internal carotid artery. The natural history is ambiguous but is typically a series of gradual developments. More frequently observed in children, where it constitutes ≥6% of strokes, it is also a well-acknowledged cause of stroke in adults. Previously regarded as a condition of Asian nations[2], it is now being reported more from India and Western countries as well. MMD is a distinct type of vasculopathy defined by features of both occlusive and proliferative disease.

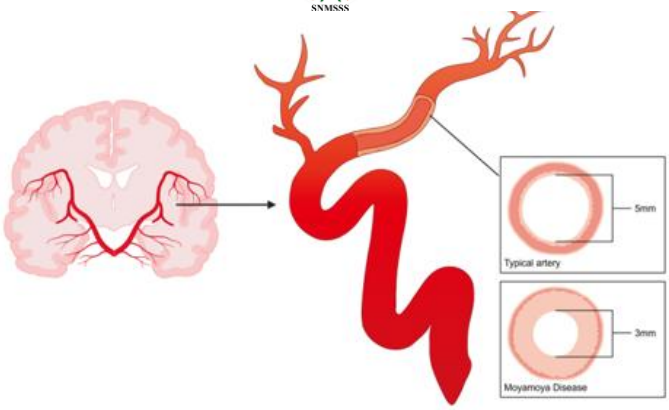

Figure 7.1: Variation in diameter of Artery.

Progressive narrowing of the terminal anterior cerebral arteries (ACAs), internal carotid artery (ICA) and leads to the compensatory development of basal perforator vessels[3]. The moyamoya vessels are arteries that run through the thalamus and basal ganglia, supplying collateral blood flow to areas of the brain with reduced perfusion that are located further away from the narrowed vessels. The compensatory system frequently malfunctions, leading to repeated strokes, whether hemorrhagic or ischemic. The precise pathogenesis and natural history remain uncertain, but they typically develop gradually. Existing evidence indicates that surgical treatment offers considerable relief from symptoms[4].

STAGES OF MOYAMOYA DISEASE[5]:

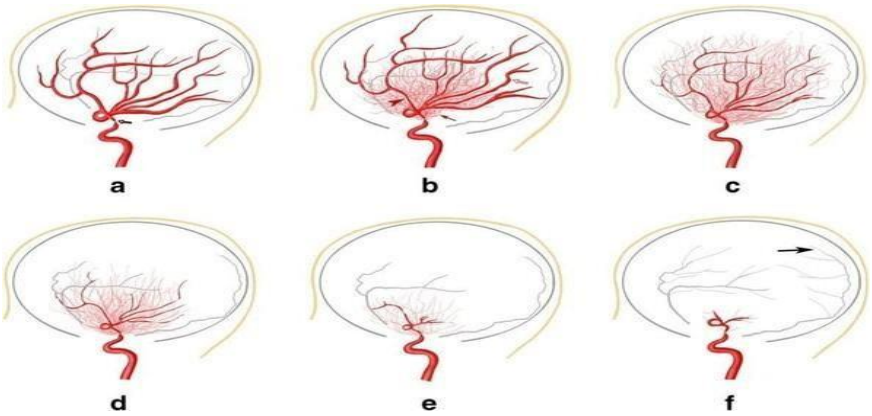

Figure 7.2: Stages of moyamoya disease

Stage a: *Constriction of carotid bifurcation* - The angiographic examination shows that only the distal segment of the internal carotid artery is narrowed.

Stage b: *Onset and manifestation of basal moyamoya* - Angiographic evaluation shows narrowing of whole terminal divisions of the IICA.

Stage c: *Basal moyamoya intensification* - The angiographic examination reveals enhanced deep moyamoya vessels. MRA conducted in this phase exhibits a "cloud of smoke" appearance.

Stage d : *Minimisation of moyamoya basal* - Lors de l'examen angiographique, les vaisseaux moyamoya profonds commencent à régresser tandis que des collatérales transdurales commencent à apparaître.

Stage e: *Decrease of moyamoya* - The angiographic examination shows ongoing reversion of deep moyamoya vessels and the advancement of trans-dural collateral vessels.

Stage f: *Moyamoya's disappearance* - The angiographic examination shows that deep moyamoya vessels are gone, with total occlusion of the ICA.

SYMPTOMS[6]:

Moyamoya disease results in various signs in both adults and children. In children, the initial indication is often a recurring transient ischemic attack (TIA). Adults can also have these symptoms, along with hemorrhage in the brain, referred to as a hemorrhagic stroke. The bleeding occurs due to the rupture of collateral blood vessels in the brain.

Signs of MMD associated with decreased cerebral blood flow:

Migraine, convulsions, fatigue, loss of sensation or inability to move face, arm, or leg muscles. This usually occurs on 1 side of the physique, difficulties with image, issues with dialogue or comprehending others, referred to as cognitive and aphasia delay, Bowel and bladder disturbances. Daily Management Techniques: Balanced Nutrition, Proper Hydration, Exercise, Refrain from Smoking and Drinking, Stress Control, Consistent Sleep Schedule.

CAUSES[6]:

Exercise, crying, coughing, straining, or a fever can trigger these symptoms. Researchers continue to investigate the causes of Moyamoya disease. Some think that a faulty, inherited blood vessel might lead to the disease. The cause might also be connected to Down syndrome and sickle cell disease.

Genetic disorders and its association: Neurofibromatosis type 1 (Association).

Acquired conditions: Radiation therapy to the head or neck, chronic meningitis, Tumor at the base of the skull, Atherosclerosis of the arteries at the skull base, Arteriosclerosis, Cerebral vasculitis.

DIAGNOSIS[7, 8]:

Moyamoya disease is typically identified by a physician who focuses on brain disorders, known as a neurologist. The neurologist examines the symptoms as well as medical and family history. The neurologist might also conduct a physical examination. Typically, multiple tests are required to identify moyamoya disease and its associated conditions.

1. Imaging using magnetic resonance: This diagnostic procedure is also referred to as an MRI. An MRI employs strong magnets and radio frequencies to produce intricate images of the brain. A healthcare provider might administer a dye into a blood vessel to visualize the arteries and veins and emphasize blood flow. This kind of examination is referred to as a magnetic resonance angiogram. A perfusion MRI might be recommended by neurologist. This kind of imaging can assess the volume of blood, flowing through the vessels. It can indicate if there is a reduction in blood flow to the brain.

2. Computed Tomography scan: The diagnostic procedure employs multiple X-rays to produce a comprehensive image of the brain. A medical expert might administer a dye into a blood vessel to enhance the visibility of blood circulation in your veins and arteries. This is referred to as a CT angiogram. This examination cannot identify the initial phases of moyamoya disease. However, it could aid in identifying issues with blood vessels

3. Brain blood vessel imaging In a cerebral angiogram, a medical specialist places a narrow, elongated tube known as a catheter into a blood vessel located in the groin. The medical management provide a subsequently utilizes X-ray imaging to direct the catheter to the brain and injects dye via the catheter into the blood vessels of the brain. Dye takes the character of the blood vessels, enhancing their visibility during X-ray imaging.

4. Positron emission tomography scan (PET) or single-photon emission computed tomography (SPECT): During these test, a minimal dose of a harmless radioactive substance is vaccinated into the patients. PET offers visual representations of brain function and SPECT assesses cerebral blood flow.

5. EEG (electroencephalogram): It observes the brain's electrical activity through small metal discs, known as electrodes, placed on the scalp. Children suffering from moyamoya disease frequently show EEG results that fall outside of the usual range.

TREATMENT[9,10]:

1. Chemotherapy: Medications might be recommended to alleviate symptoms, lower the likelihood of a stroke, or assist in controlling seizures. Medications might consist of:

• Anticoagulants: The moyamoya disease were diagnosed with mild or no symptoms, blood thinners are generally advised. The neurologist might suggest taking aspirin or other anticoagulant to prevent stroke.

• Calcium channel antagonists: Also referred to as calcium blockers, this class of drugs might assist in controlling headaches. It might also aid in diminishing symptoms associated with transient ischemic attacks. Calcium channel blockers can aid in controlling blood pressure, which may assist in preventing blood vessel harm in individuals with moyamoya disease.

• Medications for seizure control: These medications may be beneficial for individuals who have experienced seizures.

2. Surgery: In revascularization procedures, surgeons create bypasses around obstructed arteries. They achieve this by linking blood vessels from the exterior of the skull to the interior to aid in restoring blood circulation to your brain. This can encompass both direct and indirect revascularization techniques. Or it might involve a mix of both.

• Procedures for direct revascularization: The neurosurgeons connect the scalp artery straight to a brain artery using stitches. This procedure from the superficial temporal artery to the middle cerebral artery is also known as bypass surgery. The process promptly enhances blood circulation to the brain.

• Non-direct techniques for revascularization: In indirect revascularization, the objective is to enhance blood circulation to the brain progressively. In adults receiving treatment at high-volume surgical centers, indirect revascularization is nearly always used alongside direct revascularization.

PREVENTION

Moyamoya disease cannot be prevented. However, strokes and other problems can be avoided with moyamoya therapies.

RESEARCH EFFORT[11]:

Research will enhance our comprehension of moyamoya disease and result in advancements in diagnosis, treatment, and prevention. Numerous studies in progress focus on examining particular genes (like ring finger protein 213, or RNF213) for potential links to moyamoya, assessing imaging methods to enhance disease progression monitoring, and refining prediction models for patients facing revascularisation surgery. There is a lot to discover from the firsthand experiences of moyamoya patients following a successful operation, and performing routine tests or checkups will yield essential information to improve treatment methods.

CONCLUSION:

Moyamoya disease is a diagnosis that occurs by ruling out other conditions. Much more prevalent is the moyamoya syndrome, which arises as a result of the existence of additional risk factors. In addition, it is essential that the treating physician manages any of these risk factors present. In scenarios such as sickle cell anemia, a frequent cause of the syndrome, consistent blood transfusions significantly decrease the chances of a life-threatening stroke. The rare moyamoya disease is elusive, has a high mortality rate, and requires further research to clarify its etiopathogenesis. Even though treatment options, mainly neurosurgical, are advancing quickly at present, over time, one can only anticipate an improvement in outcomes.

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Chapter - 8

A Comprehensive Assessment of Brain Tumor

Nitin. S

ABSTRACT:

Brain tumors rank among the top reasons for mortality. Brain tumor is an irregular mass found in any tissue inside the skull, including the brain, cranial nerves. meninges, glands (pituitary and pineal). The unusual development of brain tissue can significantly impair brain function and ultimately affect the whole body. It is linked to a gradual decline in emotional and mental well-being, leading to a diminished value of lifetime. A brain tumor arises from the brain referred to as metastatic. Benign tumors are non-cancerous, typically confined, grow slowly and possess a uniform structure. Malignant or cancerous tumors, in contrast, have a non-homogeneous structure and exhibit invasive growth alongside a rapid proliferation rate. In this review, we outline various types and diagnostic methods for assessing contemporary treatment choices and managing life post-diagnosis, providing knowledge and fostering a sense of optimism.

Keywords: Brain tumors, Identification, Progress and evolving field

INTRODUCTION:

Human brain, acting as the center for controlling all body organs, is a refined organ that allows persons to adjust and endure different ecological conditions[1]. The human brain allows individuals to eloquent themselves verbally, convey emotions, perform tasks, and ideas[2]. The 3 primary tissue elements of the human brain are cerebrospinal fluid, gray matter and white matter. The gray matter controls brain function and consists of neurons and glial cells[3]. The cerebral cortex is linked to various brain regions via white matter fibers that consist of multiple myelinated axons. The corpus callosum, a large bundle of white matter fibers, links to the both hemispheres of the brain[4]. A brain tumor is an uncontrolled and abnormal growth of brain cells. Any unforeseen change can impact human performance, as the human skull is a stiff and capacity limited framework, contingent on the brain region affected.

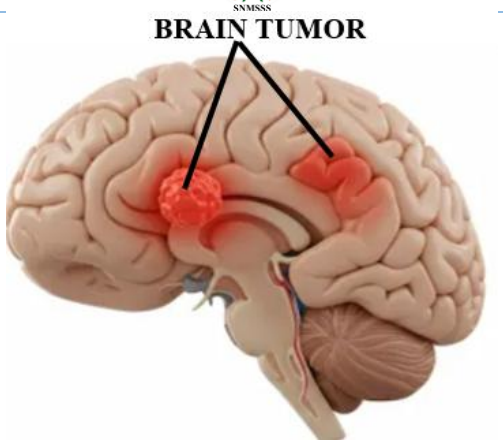

Figure 8.1. Showing tumor cells in the Brain

Moreover, it could extend to other organs, increasing the risk to human functions. Timely cancer identification enables the formulation of active usage strategies, which is vital for the pharm industry[5]. Cancer is difficult to treat, and survival rates drastically decline, if it metastasizes to adjacent cells. Certainly, countless patient might be saved if cancer were identified at its initial phase through rapid and cost-effective diagnostic techniques. Diagnosis of brain cancer can involve both invasive and noninvasive methods. During a biopsy, a lesion sample is taken for analysis by making an incision[6]. Pathologists observe numerous cellular physiognomies of the tumor sample with help of microscope to confirm malignancy, which is regarded as the gold standard in cancer diagnosis.

EFFECT OF BRAIN TUMORS[7]:

A brain tumor can have devastating consequences that affect a person's mental, emotional, and physical capacities. The symptoms, which impact general well-being, can range from slight changes to severe neurological abnormalities. Disabilities with movement, speech, memory, focus, emotions, and character may arise from the illness and its treatments. Patients often require long-term care and extensive rehabilitation.

CAUSES[8]:

- *Proto-oncogenes:* These genes usually promote the division and growth of cells. When changed, they can become oncogenes, which are like accelerator pedals stuck to the ground and cause uncontrolled cell division.

- *Tumor Suppressor Genes:* These genes typically manage cell growth, fix DNA damage, and initiate programmed cell death (apoptosis) when cells turn abnormal. When altered or deactivated, they resemble defective brakes, permitting abnormal cells to proliferate and replicate uncontrollably.

- *DNA Repair Genes:* Changes in these genes can result in the build-up of additional detrimental mutations, raising the risk of cancer.

Numerous factors have been examined, yet scientific proof does not reliably indicate a direct causal connection: Cell Phone Usage, Head Trauma, Infections (Epstein-Barr virus - EBV), Environmental Toxins/Pesticides.

CATEGORIES[8]:

Table: 8.1 Types and Origination

Types	Originate	Information
Gliomas	Glial cells	Neutron-supporting and protecting cells
Astrocytomas	Astrocytes	Most common and ranging from mild to extremely violent
Oligodendrogliomas	oligodendrocytes	Develop slowly or rapidly.
Ependymomas	Neoplasms - ependymal cells	Line the ventricles of the brain
Meningiomas	Meninges	Membranes that surround the brain for protection.
Pituitary adenomas	Pituitary gland that	Influence hormone secretion
Medulloblastomas	Cerebellum	Primarily affecting children.
Colloid cysts	Brain's third ventricle	Cause the brain's pressure to build up dangerously.

SYMPTOMS[9]:

Symptoms of a brain tumor differ significantly based on the tumor's size, position, growth speed, and if it's leading to swelling or heightened pressure inside the skull.

Common Symptoms (Due to Increased Intracranial Pressure): Headaches, Nausea and Vomiting, Changes in Mental State/Personality, Vision Problems.

Specific Symptoms:

- Frontal Lobe: Changes in Personality and Behavior, Weakness or Paralysis, Difficulty with Speech, Cognitive Dysfunction.
- Temporal Lobe: Convulsions, Memory Difficulties, Communication and Language Challenges, Hearing Hallucinations
- Parietal Lobe: Sensory Issues, Challenges with Writing or Math
- Occipital Lobe: Vision Problems
- Cerebellum: Issues with Coordination and Balance, Speech Challenges, Nausea and Vomiting.
- Brainstem: Visual Disturbances (Diplopia), Challenges with Swallowing (Dysphagia) or Speaking, Alterations in Pulse Rate, Respiration, or Awareness.
- Hormonal Imbalances: Depending on the hormones affected
- Vision Problems: Resulting from stress on the optic chiasm (the point where optic nerves intersect).

DIAGNOSIS METHOD[9]:

Identifying a brain tumor necessitates a thorough strategy that includes various clinical evaluations and sophisticated imaging methods.

- Neurological Assessment: A doctor will examine several aspects of nerve activity, the brain, and the spinal cord. Vision, balance and coordination, strength, sensation, cognitive function, and reflexes are all evaluated in this process.
- Imaging Procedures: MRI-Magnetic Resonance Imaging, MRS-Magnetic Resonance Spectroscopy, CT- Computed Tomography, PET - Positron Emission Tomography and Cerebral Angiography are examples of imaging procedures.
- Biopsy: A neuropathologist must obtain a tiny sample of the tumor tissue for microscopic examination.

This permits:

- Verifying the existence of a tumor.
- Assessing whether it is noncancerous or cancerous.
- Determining the particular kind of tumor.
- Evaluating the tumor according to its level of aggressiveness.

- Conducting molecular examinations to detect particular genetic mutations or biomarkers that can direct targeted treatments.

Biopsy Types: Stereotactic Biopsy, Open Biopsy (Craniotomy), Lumbar Puncture (Spinal Tap).

TREATMENT[10-11]:

The main treatment modalities include:

1. Surgery: The main objective of brain tumor surgery is to excise as much of the tumor safely as feasible, avoiding the introduction of new neurological deficits. Total removal (gross total resection) is often preferable, particularly for benign tumors, since it can lead to a cure. For cancerous tumors, complete safe removal can enhance results and increase the efficacy of additional therapies.

2. Techniques:

Craniotomy: The most frequent surgical operation, which entails temporarily taking out a part of the skull to reach the brain.

Awake Craniotomy: In certain instances, patients remain conscious during segments of the procedure, enabling the surgeon to assess neurological functions (e.g., speech, movement) in real-time, reducing harm to essential brain regions.

Neuro-navigation and Intraoperative Imaging: Surgeons employ advanced technologies to create a "GPS" for the brain, guiding them precisely to the tumor and helping avoid critical structures.

Minimally Invasive Techniques: Endoscopic techniques may be applied for certain tumors, particularly in the pituitary area or ventricles.

3. Therapies:

- Radiation Therapy: Utilises high-energy radiation to eradicate cancer cells and reduce tumors. It is repeatedly utilised post-surgery to eliminate residual tumor cells or as an initial treatment when surgery cannot be performed.

Types: EBRT - Proton Beam Therapy, External Beam Radiation Therapy, SRS - Stereotactic Radiosurgery, FSRT - Fractionated Stereotactic Radiotherapy.

- Chemotherapy: Temozolomide is a frequently prescribed oral chemotherapy medication for glioblastoma and various gliomas.
- Targeted Therapy: Targeted therapies obstruct growth factor receptors on cancer cells or impede angiogenesis.

- Immunotherapy: Acts by inhibiting proteins that hinder immune cells from targeting cancer cells.

4. Symptom Management: Drugs such as corticosteroids, anti-seizure drugs, and painkillers are essential for alleviating symptoms and enhancing life quality.

5. Rehabilitation: Physical therapy, occupational therapy, speech therapy, and neuro-psychological rehabilitation are essential for assisting patients in regaining lost abilities and enhancing daily living skills.

6. Palliative Care: Concentrates on alleviating the symptoms and stress associated with a severe illness, intending to enhance patient's and their family's quality of life.

RECENT ADVANCES IN BRAIN TUMOR RESEARCH[12-13]:

- **Precision Diagnosis:** Contemporary diagnosis increasingly relies on molecular analysis of tumour specimens. This involves examining the tumour's DNA, RNA, and proteins to identify specific genetic mutations and epigenetic alterations. This allows for:
- **Identification of Actionable Targets:** These molecular findings identify particular "targets" that medications can obstruct.
- **Liquid Biopsies:** This non-invasive method is becoming increasingly popular. It includes examining body fluids for circulating tumor DNA or circulating tumor cells released by the tumor.
- **Checkpoint Inhibitors:** The drug inhibit proteins on immune cells that tumors exploit to "conceal" themselves from the immune system, enabling the immune cells to identify and assault the cancer. Although progress has been restricted in certain prevalent brain tumors such as glioblastoma, investigations persist.
- **Cancer Vaccines:** Vaccines tailored to individuals are being created to educate the immune system to identify and target particular proteins produced by cancer cells. Initial clinical trials are investigating this for aggressive brain tumors.
- **Fluorescence-Guided Surgery:** , patients are given a particular dye (such as 5-ALA) that, when exposed to certain light, makes tumor cells glow. This helps surgeons distinguish between tumor and healthy brain tissue more precisely when removing the tumor.
- **Neuro-navigation and Robotics:** Computer-assisted navigation systems and robotic arms increase surgical precision, allowing for less invasive procedures and accurate tumor targeting.

EMERGING RESEARCH AREAS:

- **Artificial Intelligence (AI):** AI is used to assess complex genetic, pathological, and imaging data in order to predict treatment response, help with diagnosis, and identify new therapeutic targets..
- **Gene Therapy:** Research is looking into ways to introduce new genes into cancer cells to increase their susceptibility to therapies or give them the ability to produce compounds that are harmful to themselves.
- **Oncolytic Virus:** Researchers are investigating genetically modified viruses known as oncolytic virus which target and eliminate cancer cells only, leaving healthy cells intact.

The above developments represent a change toward more individualized, minimally invasive, and successful brain tumor treatments, giving patients new hope.

CONCLUSION:

Malignant brain tumors encompass various tumor types, each requiring specific chemotherapy protocols tailored to the pathological diagnosis. Frequently, both healthcare providers and the general public view chemotherapy treatments for patients with brain tumors as ineffective. Recent advancements in chemotherapy studies have offered patients novel therapeutic alternatives that could increase their chances of survival and enhance their quality of life, while minimizing possible toxicities. Additionally, the continuous focus on cytotoxic and small-molecule drugs provides new hopefulness that these cutting-edge treatments will improve results for individuals facing malignant brain tumors. Timely and precise identification of brain tumors is crucial for effective therapy and treatment planning. Recently, many research studies have been carried out on the diagnosis and treatment of brain tumors. The information from the new imaging methods MRI, CT scan, and PET scan is anticipated to aid in early detection and provide more precise outcomes

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Chapter - 9

Alzheimer's Disease: A Brief Analysis

Preethi. V

ABSTRACT:

Alzheimer's disease is the leading origin of dementia, representing 60-70% of all occasions. It is a neurological disease that degenerates with time and is characterized by cognitive decline, memory loss and difficulties with everyday tasks. It affects a great number of people worldwide. Communities and healthcare systems are severely impacted financially by Alzheimer's disease. Understanding the pathophysiology of Alzheimer's disease, improving diagnostic methods, and developing efficient treatments are essential given its increasing prevalence. This thorough analysis investigates the complex processes associated with Alzheimer's disease, assesses the present situation of diagnostic methods, and evaluates new treatment approaches. This review intends to enhance the expanding understanding of this devastating disease by uncovering the complexities of Alzheimer's disease

Keywords: Alzheimer's Disease, Causes, Diagnostics, Therapeutics.

INTRODUCTION:

Alzheimer's disease (AD) is a advanced neurodegenerative condition that mainly impacts memory, understanding and behavior[1]. In older adults, it is marked by the progressive decline of cognitive abilities resulting from the death of brain cells. It typically occurs in individuals older than 65 years[2]. Because of an older population, each year, we notice a rising number of impacted patients. More than 50 million individuals are expected to have AD dementia, and this number is likely to increase three-fold by 2050[3].

At the biological level, AD is linked to the accumulation of two irregular protein formations in the brain-amyloid plaques and neurofibrillary tangles composed of tau protein. These alterations interfere with neuron communication and ultimately result in cell death[4]. While the precise cause of Alzheimer's remains unclear, it is understood that a mix of genomic predispositions and ecological influences play a role in its onset. At present, there is no remedy, but therapies exist to alleviate symptoms and enhance quality of life. Many clinical trials of monotherapy did not prevent disease progression or enhance symptoms relative to sample[5]. Research shows that the pathological changes associated with AD begin many years before the disease becomes evident.

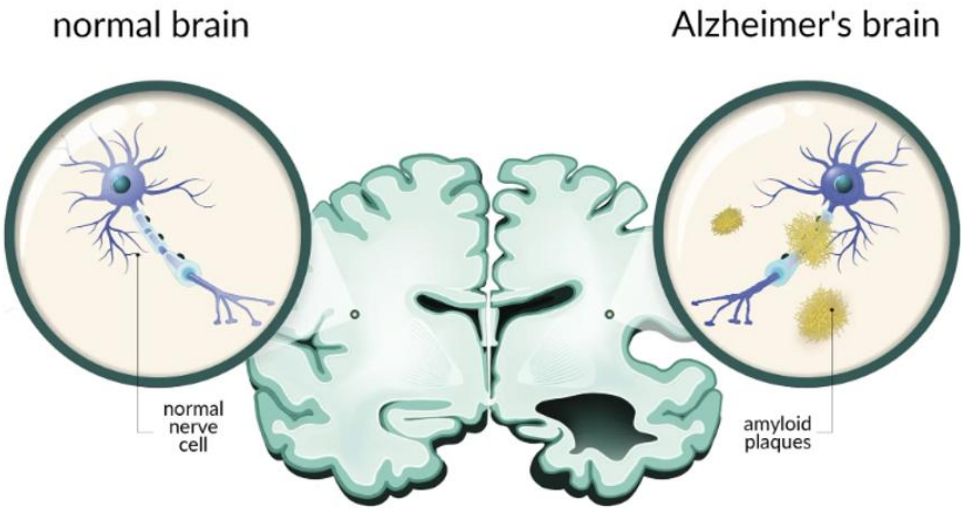

Figure 9.1. Difference between the normal and Alzheimer's Brain

CAUSES[6]:

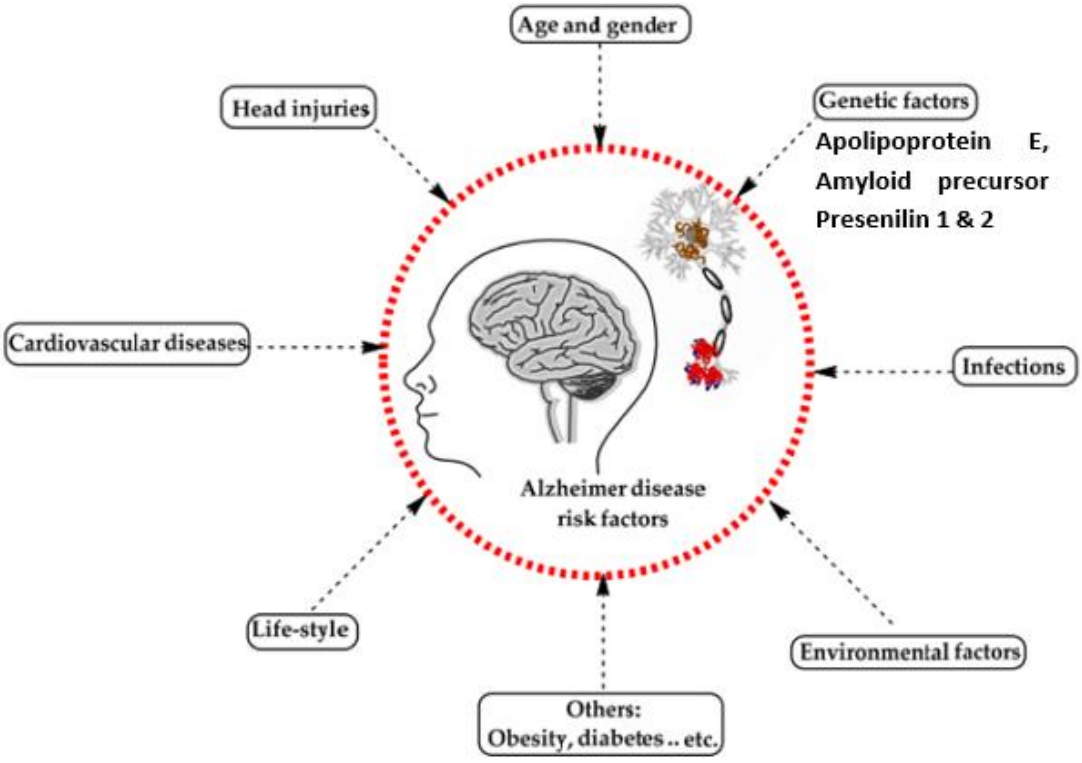

Figure 9.2. Causes of Alzheimer's Brain

SYMPTOMS[7]: The symptoms are not consistent and can differ from individual to individual,

but they generally adhere to a recognizable pattern corresponding with the stages of the disease are mild (early), moderate (middle), and severe (late). These symptoms indicate the foundational injury and demise of brain cells, especially in areas related to memory, reasoning, language, and behavior.

Initial Symptoms (Mild):

- During the initial phase, people might continue to manage on their own but start to observe slight cognitive challenges.
- These may be confused with typical aging but are generally more enduring and advancing.

Symptoms include: Loss of short-term memory, Challenges in planning or problem-solving, Disorientation about location or time, Communication issues, Misplacing objects.

Middle-Stage (Moderate) Symptoms:

- As the illness worsens, symptoms get worse and interfere with daily activities.
- This stage could last for several years.

Symptoms include: Increased memory loss, increased communication difficulties, behavioral and emotional changes, sleep patterns that vary, becoming lost and confused, difficulties with personal care tasks, and recurring behaviors are some of the symptoms.

Late-Stage (Severe) Symptoms:

- Alzheimer's significantly impairs mental and physical health in its latter stages.
- People become fully reliant on their caregivers.

Symptoms include: Impaired communication, Significant memory loss, Lack of mobility, Incontinence, Greater susceptibility to infections.

Neuropsychiatric / Behavioral and Psychological Symptoms of Dementia :

- Hallucinations and delusions, Aggression and irritability, Depression and apathy, Disturbances in sleep.
- These symptoms arise from injury to different regions of the brain, such as the frontal and temporal lobes

TREATMENT[10]:

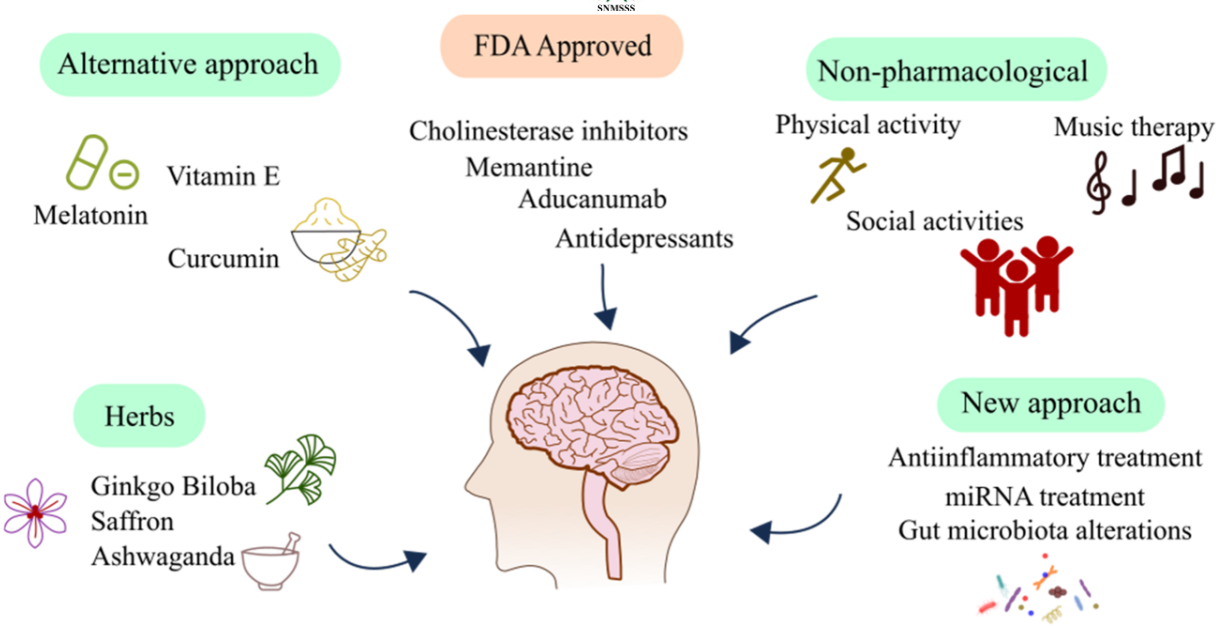

Figure 9.2. Treatment of Alzheimer's Brain

DIAGNOSIS METHODS[8,9]:

1. Brain Imaging Methods: Neuroimaging is essential for identifying brain alterations typical of Alzheimer's and excluding other disorders (e.g., tumors, strokes).
    - Magnetic Resonance Imaging (MRI) - Identifies brain shrinkage, primarily in the hippocampus, a region crucial for memory, and eliminates the possibility of strokes, tumors, or other structural irregularities.
    - Computed Tomography (CT) Scan - Effective for detecting significant structural alterations in the brain and offers less detail than MRI but is more available in certain environments.
    - Positron Emission Tomography (PET) Scan – Assesses brain metabolism; reduced glucose absorption in specific brain regions indicates Alzheimer's disease.

2 PET scans for amyloid and tau: Identify protein buildups associated with AD laboratory examinations.

3: Genetic testing: Detection of the APOE might be utilised in specific instances or for research, especially in early-onset or hereditary Alzheimer's.

4. Biomarkers in Cerebrospinal Fluid (CSF): Lumbar puncture can be performed to evaluate CSF concentrations of:

- Amyloid-beta 42 (Aβ42) – typically reduced in AD,
- Total tau (t-tau) – elevated due to neurodegenerative processes,
- Phosphorylated tau (p-tau) – linked to tau-related pathology.

CSF examination can enhance diagnostic precision, particularly in early or ambiguous situations.

5. Developing Methods:

- Plasma biomarkers are exhibiting potential as non-invasive methods for future diagnosis.
- AI and machine learning are being created to forecast Alzheimer's disease through MRI and cognitive assessment information.

RECENT RESEARCH[11]:

Recent breakthroughs in science have concentrated on deciphering the fundamental causes of Alzheimer's disease and creating treatments to slow down or avert its advancement. Neuroscientists and medical experts around the world are now researching a wide range of topics.

1. **Therapies Using Anti-Amyloid Antibodies:**

   Donanemab: During phase 3 clinical trials, this prospective drug, which targets amyloid, showed encouraging results in postponing the functional deterioration and development of cognitive in early AD (2023).

2. **Treatments Focused on Tau Proteins:**

   Researchers are developing drugs that stop tau tangles, a key feature of Alzheimer's disease, from forming or spreading. Anti-tau antibodies and tiny molecules intended to prevent tau clumping in the brain are being tested in early studies**.**

3. **Blood-Derived Biomarkers for Early Identification:**

   New studies have discovered blood tests that can detect abnormal tau and amyloid-beta protein levels. These indicators may make it possible to detect Alzheimer's disease early and non-invasively, years before symptoms appear, allowing for early treatments.

CONCLUSION:

Alzheimer's disease is a long-term neurological illness that causes severe memory loss, cognitive decline, and behavioural changes. Along with genetic, lifestyle, and environmental factors, the buildup of tau tangles and amyloid plaques in the brain is the main cause. Millions of people worldwide are still affected by the sickness, which has numerous medical and societal repercussions because age is still the main risk factor. Although there is currently no cure, recent research has produced promising treatments that can slow down the advance of the illness and

recover the quality of life. Managing Alzheimer's disease requires early detection, support for caregivers, and adoption of good lifestyle choices. In order to advance toward successful preventative and long-lasting remedies, ongoing research and awareness are crucial.

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Chapter - 10

An Assessment on Pancreatic Cancer

ABSTRACT:

Pancreatic cancer is record as the aggressive and lethal types of cancer, originating from the pancreas's tissues, an organ situated behind the stomach that is crucial for digestion and blood sugar control. Despite being less common than some other cancers, it is one of the foremost reasons of cancer-related demises globally due to its rapid progression, late discovery, and resistance to many common treatments. Since symptoms usually appear only after the disease has progressed, most instances are discovered later. Pancreatic cancer is a significant challenge in oncology because to limited treatment options and poor survival rates, underscoring the urgent need for improved medicines, early detection methods, and public education. With a focus on approved immunotherapies and diagnostic technologies, this review seeks to highlight modern approaches in clinical practice.

Keywords: Pancreatic cancer, Causes, Diagnosis, Treatment.

INTRODUCTION:

Pancreatic cancer (PC) is a formed as a cluster of cells in the pancreas which is located behind the bottom section of the stomach[1]. It produces enzymes that assist in food digestion and hormones that regulate blood sugar levels. Pancreatic ductal adenocarcinoma (PDAC) is the most common type of pancreatic cancer[2]. This form originates in the cells that line the conduits transporting digestive enzymes from the pancreas. PC represents about 2.1% of all cancer cases and close to 3.4% of cancer-related fatalities in India[3].

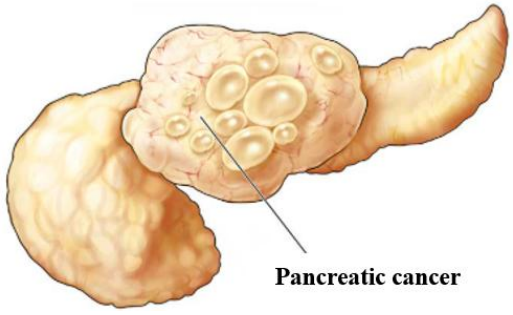

Figure 10.1. Cancer in the Pancreas

In India, PC is ranked 18th in terms of death and 24th with 10,860 (1.03%) new cases. Despite being an uncommon location, PC is a major cause of death worldwide, and information is scarce regarding the epidemiology of PC[4]. It is predicted that, as a result of the shifting lifestyle

worldwide and in India, the incidence of PC will rise in the upcoming years. PC is elevated in the elderly demographic (over 50% in ages 65-75) and is most prevalent in the Northeastern regions of India[5]. Risk factors consist of smoking, excessive alcohol intake, non-vegetarian eating habits (adaptable) and age, ethnicity, and genetic predispositions s (non- adaptable) threat factors.

TYPES OF PANCREATIC CANCER[6]:

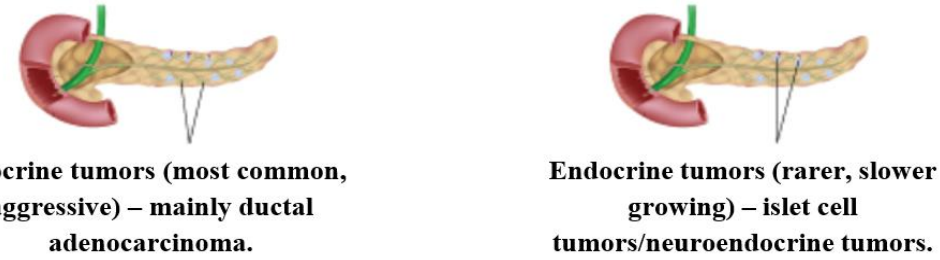

Figure 10.2. Types of Pancreatic Cancer

CAUSES[7]:

Figure 10.3. Causes of Pancreatic Cancer

SYMPTOMS[8]:

The rise in PC cases is alarming, as it primarily reflects shifting lifestyle factors, which are evolving quickly in India, similar to other regions worldwide. Pancreatic cancer is frequently untreatable when identified at a late stage, as symptoms emerge only during advanced phases after it has metastasised to other organs (liver, lungs, and peritoneum). A limited proportion (approximately 10–20%) of instances can be treated through surgical excision (such as the Whipple procedure) when identified early. In advanced cases, the typical survival duration post-

diagnosis is frequently under a year; the 5-year survival rate is still quite minimal (10%)[6].

Figure 10.4. Symptoms of Pancreatic Cancer

DIAGNOSIS[8]:

Figure 10.5. Diagnosis of Pancreatic Cancer

PREVENTION[9]:

Figure 10.6. Prevention of Pancreatic Cancer

TREATMENT[10] :

Management of pancreatic cancer generally includes a mix of surgery, chemotherapy, and radiation treatment. Only around 20% of cases, however, are eligible for surgery because

most are discovered when the disease has spread. In advanced circumstances, palliative care is essential for reducing symptoms and improving quality of life.

1. Surgery - Biopsy:

EUS or CT-guided fine-needle aspiration (FNA) - Confirms the existence of cancer using histology and cytology. If imaging clearly indicates cancer, it may not always be done before surgery.

2. Radiation therapy:

The second treatment option for pancreatic cancer is radiation therapy, which is often used in conjunction with chemotherapy and plays a supportive but essential function. Eliminating cancer cells is its main goal, whether it be to decrease the risk of recurrence after surgery, control tumor growth in cases when surgery is not possible, or shrink the size of the tumor before surgery.

3. Oncology Medications:

Gemcitabine, 5-Fluorouracil (5-FU), Capecitabine, Irinotecan, Oxaliplatin, Folfirinox, Nab-paclitaxel (Abraxane), and Cisplatin.

4. Targeted Treatment Medications:

Table: 10.1 Targeted Treatment Medications

Erlotinib	**Blocks the epidermal growth factor receptor**
Olaparib - (ADP-ribose) polymerase blocker	Maintenance treatment for individuals with BRCA-mutated metastatic pancreatic adenocarcinoma.
KRAS Inhibitors (Adagrasib and sotorasib)	Target pancreatic cancers harboring the KRAS G12C mutation.
Pembrolizumab	targeting programmed cell death protein-1 utilized for advanced pancreatic cancers exhibiting DNA repair deficiencies

RECENT ADVANCES IN PANCREATIC CANCER[11]:

1. Molecular and Genetic Profiling:

- Using genetic and molecular profiling to customize treatment to meet the needs of each patient is a significant breakthrough.
- PARP medicines like olaparib, which target specific flaws in cancer cells, are now beneficial for people with BRCA1/2 mutations or deficits in DNA repair.
- It is currently recommended that all patients with pancreatic cancer get molecular testing, not only those with a family history.

2. Liquid Biopsies and Early Detection:
   - A non-invasive method for detecting pancreatic cancer early and monitoring the effectiveness of treatment is the analysis of circulating tumor DNA (ctDNA) or exosomes in the bloodstream.
   - These technologies have the potential to identify the disease at an earlier, more controllable stage, despite the fact that they are still in the development stage.
3. Improved Chemotherapy Protocols:
   - The field of chemotherapy continues to progress.
   - Due to its increased efficacy, the FOLFIRINOX treatment has become the standard for eligible patients, and gemcitabine and nab-paclitaxel combinations are producing better outcomes with tolerable side effects.
   - Additionally, flexible chemotherapy regimens based on tumor response are being evaluated.
4. Surgical Method:
   - Less invasive surgical techniques and better preoperative planning strategies using cutting-edge imaging have improved surgical approaches.
   - Neoadjuvant therapy is becoming more and more common, improving resectability in borderline cases.
   - Stereotactic Body Radiation Therapy (SBRT) is a type of radiotherapy that provides high-dose, tailored treatment with fewer adverse effects.
5. Organoid Models and Tailored Drug Evaluation:
   - To monitor drug reactions as they happen, organoids made from patient tissue-miniature lab-cultivated copies of tumors-are used.
   - This increases the likelihood of treatment success by enabling highly customized therapy options.

CONCLUSION:

In conclusion, due to its covert growth, delayed detection, and limited treatment options, pancreatic cancer is considered to be among the most aggressive malignancies. The intricacy of the disorder makes prevention and therapy particularly challenging, even though genetics, lifestyle, and environmental factors all have an impact on its beginnings. However, patients' prognoses are beginning to change as a result of the growing use of molecular profiling, liquid biopsies, and cutting-edge therapies including immunotherapy and targeted drugs. The best way to improve survival rates is still early detection, which highlights the need of raising awareness, screening high-risk groups, and making lifestyle adjustments like quitting smoking and maintaining a healthy weight. While significant advancements are still required, the consistent flow of scientific breakthroughs provides optimism that pancreatic cancer, once seen as almost untreatable, will gradually evolve into a manageable illness in the future

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Chapter - 11

A Review of Tumor Occurring in the Spine

Suganth. M

ABSTRACT:

Tumors can develop inside or around the spinal cord or vertebral column. Spinal tumors could either be benign or cancerous. Depending on their epicenter, tumor can be categorized as intradural/extradural-extramedullary, intradural-intramedullary. Among these, extradural lesions are the utmost prevalent and are usually metastatic. 1° bone tumors of the spine account for 5 percent of all bone tumors. Almost all primary tumors are benign, while malignant tumors make up just 20 percent. In general, spinal metastases represent the most predominant malignant tumors in the spine, typically originating from primary cancers like lung, breast, and prostate. The emergence of enhanced systemic treatments resulting in better survival rates and the regular use of tomography have established metastatic spine ailment as the latest rampant in oncology. The accurate diagnosis largely depends on magnetic resonance imaging and histological verification for this tumor. This article will give a summary of the types, signs and symptoms, and treatment

Keywords: Spine tumor, Types, Causes, Diagnosis.

INTRODUCTION:

A spine tumor is an unusual tissue growth located inside or everywhere in the spinal cord and column. The tumors may be non-cancerous (benign) or cancerous (malignant), and they can be arise in the spine (primary tumors) or metastasize from other regions of the body (secondary tumors)[1]. Spine tumors can lead to considerable neurological issues, discomfort, and impairment if not identified and treated properly. Tumors in the spine can develop in various areas, such as within the spinal cord, in the surrounding protective membranes, or between the vertebrae[2]. Even small tumors may put pressure on blood vessels, nerves, or the spinal cord itself because of the limited space inside the spinal canal. Pain, weakness, tingling, numbness, or trouble moving are common symptoms of this pressure, and in extreme situations, it can even cause paralysis[3].

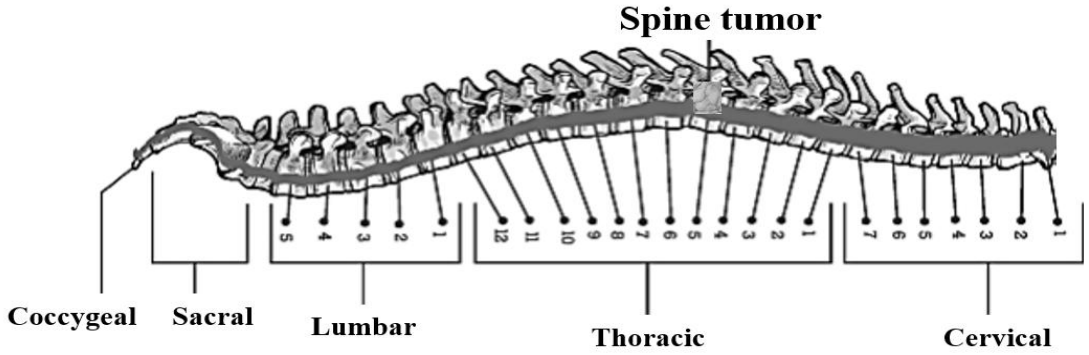

Figure 11. 1. Tumor in the thoracic bone of the spinal cord.

When examining tumors of the spine, it is essential to take into account the various tissue types surrounding the spinal column. The existence of bone, cartilage, renders and tissue (neural & meningeal) are a potential source for neoplastic alteration. Metastatic abrasions can spread from remote primary tumor locations to the spine through pathways (haematogenous/ lymphatic)[4]. Spine tumors are rare and constitute <5% of bone neoplasms, the yearly incidence of primary spine tumors ranges from 2.5 to 8.5 per 100,000 individuals. The spine is far more frequently affected by metastatic disease. Around 40-80% of spine cancer patients who pass away have bone metastases which is the most frequent site of skeletal metastasis[5].

TYPES OF SPINE TUMORS[6]: Spine tumors are classified based on their location and origin

Table 11.1. Types of Spine Tumors by Location

Intramedullary Tumors	Progress within the spinal cord.
Extramedullary - Intradural Tumors	Arise inside the duramembrane but outside the spinal canal.
Extradural Tumors (most common type)	Positioned beyond the dura mater and frequently metastatic. Engage the vertebrae (spinal bones).

Table 11.2. Types of Spine Tumors by Origin

Primary Tumors - Benign	Arise from the spinal column or spinal cord.
Secondary (Metastatic) Tumors - Malignant	Arise from tumors in other areas of the body (such as lung, breast, prostate) and metastasize to the spine.

CAUSES[7] :

The precise origin of spinal tumors is often unclear, but researchers think that a mix of genetic alterations and environmental factors contribute to their formation.

- Genetic Alterations: Modifications in a cell's DNA can lead to uncontrolled growth.
- Genetic Disorders: Uncommon hereditary conditions (neurofibromatosis or von Hippel-Lindau disease) are associated with an increased risk of spinal tumors.
- Metastasis from Different Cancers: Numerous spinal tumors are secondary, indicating they originate from cancers in other organs like the lung, breast, prostate, kidney, or colon.
- Issues with the Immune System: A compromised immune system can heighten the likelihood of abnormal cell proliferation.
- Risk factors: It include age, radiation exposure, past cancer, genetic disorders (Li-Fraumeni syndrome and tuberous sclerosis) and family history.

SYMPTOMS[7]:

- Pain in the Back or Neck: Depending on the nerve roots implicated, it may be localized or dispersed. Often, the soreness is persistent and does not improve with rest.
- Neurological Impairments: Tingling or numbness in the arms, legs, or chest; decreased muscle strength; difficulties walking or maintaining balance; and, in extreme cases, loss of control over one's bowels or bladder.
- Paralysis: The spinal cord may be compressed by advanced malignancies, resulting in partial or complete paralysis beneath the tumor.
- Deformity: The spine may become unstable or deformed as a result of vertebral tumors.

DIAGNOSIS[8]:

Diagnosing a spine tumor involves a thorough clinical evaluation and imaging studies:

Table 11.3. Types of Imaging Studies

Medical History	**Symptoms and medical background of the patient**
Physical Examination	Neurological assessment to identify sensory, motor, and reflex irregularities.
MRI scan	Comprehensive visuals of the spinal column and adjacent structures.
CT scan	Enhanced depiction of bone involvement
X-rays	To identify bone damage or spinal alignment problems.
PET Scan	Differentiate between benign and malignant tumors.
Biopsy	Identify - tumors, infections, and inflammatory disorders

TREATMENT[9]:

- Observation: For small, non-cancerous, slowly growing tumors that are asymptomatic, routine observation with intermittent MRIs might be adequate.
- Surgical methods:
  - Laminectomy: Extraction of a portion of the vertebra to decrease pressure.
  - Spinal Fusion: Stabilising the spine through the use of rods, screws, or bone grafts.
  - Microsurgery: Techniques that are minimally invasive for intricate intramedullary tumors.
- Radiation Treatment: Frequently utilised for cancerous tumors, leftover tumors after surgery, or when surgical options are impractical. Sophisticated methods such as stereotactic radiosurgery enable precise radiation delivery while causing little harm to surrounding healthy tissue.
- Chemotherapy: Used to treat certain malignant tumors, like lymphomas or cancer that has spread. For a more potent reaction, it can be combined with radiation.
- Recuperation: Extent of nerve or spinal cord damage.
- Physical rehabilitation: Goal is to increase the restoring strength, mobility, and coordination.
- Occupational therapy - It helps people get used to their daily routines.

- Psychological Support: Therapy or support groups may be necessary while coping with a cancer diagnosis, persistent pain, or disability.

Table 11.4. Types of Medications

Corticosteroids	Help reduce inflammation and edema around the spinal cord.
Pain control	Aid to decreases nerve pain, muscle relaxants, and pain relievers.
Bisphosphonates	Used to strengthen bones in cases of metastatic spine diseases.

PROGNOSIS OF SPINE TUMOR[10]:

Factors Affecting Prognosis -

- Tumor type – Benign vs malignant.
- Location – Tumors compressing critical nerves might lead to a poorer prognosis.
- Size and stage – Early detection of smaller tumors makes treatment simpler.
- Age and health of the patient: Younger and healthier patients typically recover more quickly.
- Response to treatment: While certain cancers may be resistant to chemotherapy, radiation, or surgery, others may respond well to these treatments.
- Quality of Life Considerations: While a complete recovery may not be possible, treatments usually aim to reduce pain, improve mobility, and maintain independence. Long-term health requires counseling, physical therapy, and rehabilitation.

Survival and Recovery -

- Benign tumors: Strong likelihood of complete recovery post-surgery.
- Malignant primary tumors: The outlook relies on prompt identification and effective treatment.
- Metastatic tumors: Usually linked to advanced cancer, however contemporary therapies improve comfort and survival.

Significance of Follow-up - Routine medical appointments, MRI assessments, and neurological evaluations are crucial for tracking recurrence or complications post-treatment

CONCLUSION:

Spinal column is the principal part of the axial skeleton that upholds essential organs. Metastatic tumors are the most common tumors affecting the spine. Regarding occurrence, benign bone tumors are more common than metastatic sarcomas, while primary bone sarcomas are the least common tumors affecting the spine. At times, spine tumor can be challenging to differentiate primary tumors from metastatic tumors in the spinal area. Metastatic tumors of unidentified origin are frequently found in the spine. Understanding the initial lesion is vital for the treatment protocol of metastatic spinal tumors. Consequently, in cases of primary unknown metastases, biopsy is a crucial step that influences treatment options. It is important to remember that metastatic lesions affecting the spine are components of systemic cancers. Surgical staging plays a crucial role in deciding the treatment approach. The management of metastatic tumors should focus on alleviating pain while maintaining the spine's mechanical and neurological functions. In treating primary tumors, the strategy must focus on eliminating local disease while maintaining the spine's mechanical and neurological functions. Like all oncological disease management, the diagnostic and interventional techniques for primary or metastatic spine tumors and the overall patient management should adopt a multidisciplinary strategy

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Chapter - 12

Prediction of Protein Involved in Oral Cancer Using NCBI Database

Shri Shobitha. S

ABSTRACT:

Oral cancer is one of the top ten most common cancers worldwide, characterised by delayed medical identification, unfavourable prediction, a lack of specific biomarkers for the condition, and expensive treatment. Oral cancer remains a significant worldwide health issue. With advancements in early detection and therapy, the outcome is better for early detected patients and those diagnosed at later stages, the outcome remains bleak. Oral cancer can be prevented if its risk factors and natural progression are understood and with advancement in biomedical sciences, especially dentistry, it is expected for better clinical outcomes. Nonetheless, advancements in comprehending the molecular processes of oral cancer development and the creation of novel therapeutic strategies provide renewed optimism for improving patient outcomes. This review emphasizes the recent forecasts of proteins linked to oral cancer, along with their symptoms, diagnosis, and treatment options.

Keywords: Oral Cancer, Protein, symptoms, Treatment.

INTRODUCTION:

Oral cancer, part of the head and neck cancer category, arises in the tissues of throat or mouth. Oral cancer typically affects individuals older than 40, and each year, thousands are diagnosed with this condition[1]. The prognosis and therapy depend on the diagnosis and the stage at which it is identified. Oral cancer can arise in different areas of the mouth, such as the lips, tongue, cheeks, gums, palate, and the inside of the mouth[2].

As per Globocan Data, India records the highest incidence of Oral Cancer cases in recent years. India accounts for one-third of all Oral Cancer Cases globally, representing 30% of the worldwide total. Every year, approximately 77,000 new instances of Lung Cancer arise, resulting in 52,000 fatalities, which indicates a death rate ranging from 70% to 80% for Oral Cancer. Additionally, the statistics indicate that men are at a higher risk for Oral Cancer than women[3]. Oral cancer is characterized by the unchecked proliferation of cells that gradually invades and impacts surrounding organs, while comprehending the specific type of oral cancer aids in ensuring accurate diagnosis and treatment[4].

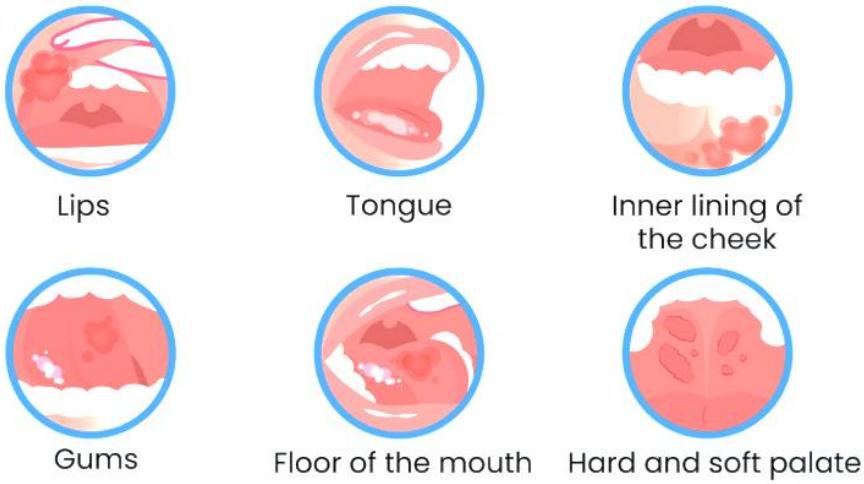

Figure 12.1. Tumor in the different parts of the Mouth.

CAUSES OF ORAL CANCER[5]:

Figure 12.2. Causes of Oral Cancer

SYMPTOMS[6]:

An ulcer on or in the lip or mouth which does not cure, loose teeth, bleeding within the mouth, a bump or growth located anyplace in the mouth, ill-fitting dentures, numbness - (lower lip, face, neck or chin), a swelling in the neck, pain or stiffness in the jaw, persistent ear pain, a raspy voice, discomfort in the tongue, challenges in chewing or swallowing, unwanted weight loss, white or red lesions in or on the mouth or lips and difficulty moving the tongue or jaw.

DIAGNOSIS OF ORAL CANCER[8,7]:

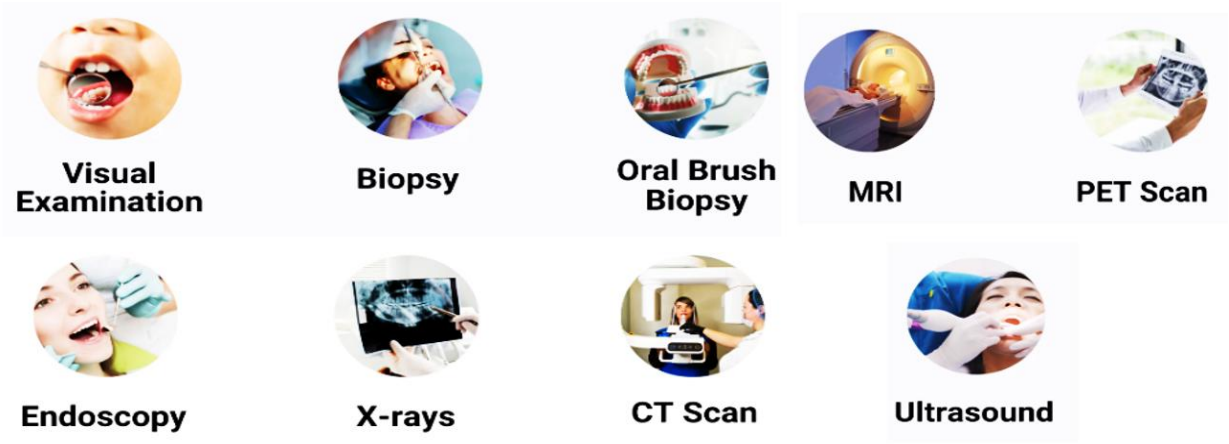

Figure 12.3. Diagnosis of Oral Cancer

TREATMENT[9]:

Management for oral cancer might include a blend of these strategies:

1. Surgery: Primary Tumor Removal, Glossectomy (Tongue Removal), Mandibulectomy (Jawbone Removal), Maxillectomy (Palate Removal), Neck Dissection, Mohs Surgery, Tracheostomy, Gastrostomy Tube Insertion, Reconstructive Surgery.
2. Radiation Treatment: External Radiation Treatment, Internal Radiation Treatment (Brachytherapy)
3. Medication-Based Therapies: Immunotherapy, Chemotherapy, and Targeted Therapy Issues: Swallowing difficulties and Speech problems,

Psychological impacts: Decreased self-esteem and social anxiety and depression.

PREVENTION STRATEGIES[11]:

Avoiding alcohol and tobacco, receiving an HPV vaccination, limiting UV radiation exposure, maintaining a healthy weight and diet, and scheduling routine dental checkups, will significantly reduce the risk of tongue cancer:

Proteins involved in oral cancer predicted by NCBI Database

Table 12.1. Proteins involved in oral cancer predicted by NCBI Database

Protein name	p53	EGFR (Epidermal Growth Factor Receptor)	Ki-67	VEGF (Vascular Endothelial Growth Factor)
Defination	Tumor suppressor protein that regulates the cell cycle	Overexpressed in oral squamous cell carcinoma	Marker of proliferation	Promotes angiogenesis to support tumor growth
Sourcs	mutations lead to uncontrolled growth Cellular tumor antigen p53	promoting cell proliferation. E3 ubiquitin-protein ligase CBL	higher levels indicate aggressive tumors.	Vascular endothelial growth factor A, long form
Gene Name	Gene: TP53 (P53)	Gene: CBL (CBL2, RNF55)	Gene: MKI67	Gene: VEGFA (VEGF)
Organism	Homo sapiens (Human)			
Amino acid no.	393 amino acids	906 amino acids	3256 amino acids	395 amino acids
Expression	Evidence at protein level			
Function	Regulates cell cycle and induces apoptosis in response to DNA damage.	Promotes cell proliferation and survival upon activation.	Indicates cellular proliferation rate.	Stimulates angiogenesis and lymphangiogenesis
GenBank	BAC16799.1	NP_005179.2	sp|P46013.2	sp|P15692.3

CONCLUSION:

Patient outcomes could be significantly improved by developing novel diagnostic and therapeutic approaches as well as by better understanding the proteins associated with oral

cancer pathogenesis. More customized treatment options are now available thanks to the development of targeted treatments and immunotherapies brought about by the use of molecular biology in clinical settings. Ongoing investigation into the molecular mechanisms, coupled through creation of innovative treatment approaches, is crucial for enhancing quality of life and death rates for individuals affected by severe illness.

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Chapter - 13

In Silico Evaluation of Gene Mutation in Progeria Syndrome Using Bioinformatics Database

Subhashini. K

ABSTRACT:

Progeria, acknowledged as Hutchinson-Gilford progeria syndrome, is an rare genetic form marked by significant early aging and rapid development of cardiovascular ailments. It rarely gets transmitted from parent to offspring. Progeria always caused due to the de novo point mutation in the lamin A gene which triggers a hidden splice donor site, resulting in a shortened mutant protein called progerin. Progeria presents a distinctive facial look, featuring flat eyes, thin lips, a narrow nose of pointed tip, a small chin and sticking out ears, alongside severe arterial hardening starting in childhood. Historically, physicians relied on physical symptoms alone to diagnose progeria, but Progeria research foundation has created a Progeria tissue and cell bank to aid in advancing research and diagnosis. This study examines the mutation's location in the gene when it is translated into protein, along with its regulation

Keywords: Progeria, Genetic disease, Lamin A, Mutation.

INTRODUCTION:

Hutchinson–Gilford Progeria Syndrome (HGPS) is an uncommon genetic condition that causes children to age significantly quicker than usual. It results from a mutation in the Lamin A (LMNA) gene located on chromosome 1, which typically generates the lamin A protein. LMNA protein complexes aid in maintaining the stability of the cell nucleus and preserving the integrity of the genomes[1]. When Lamin A mutates, it results in the creation of a defective protein known as progerin, which destabilizes the nucleus and damages DNA, ultimately causing aging effects. Consequently, impacted children exhibit symptoms such as hair loss, fragile skin, stiff joints, and premature heart disease. HGPS is categorized as an autosomal dominant disorder, with the mutation arising spontaneously. Children can be impacted even when their parents are well, and there is often no family history present. It affects 1 in 18 million children globally and is characterized by various clinical signs of early aging[2]. Children seem well at birth; however, in their early years, signs of illness emerge such as growth deficits, lagophthalmos, hair thinning, loss of subcutaneous fat, delayed and partial primary teeth emergence, unusual skin and scleroderma-like skin alterations on the lower abdomen along with fiasco to thrive. Motor,

mental, and emotional growth is unaffected in children with Progeria[3]. Abnormal physical growth may manifest as complete alopecia, a horse-riding posture, a high pitched voice, a narrowed upper thorax, subcutaneous lipodystrophy, coxa valga with potential hip dislocations, advancing joint contractures and nail dystrophy.

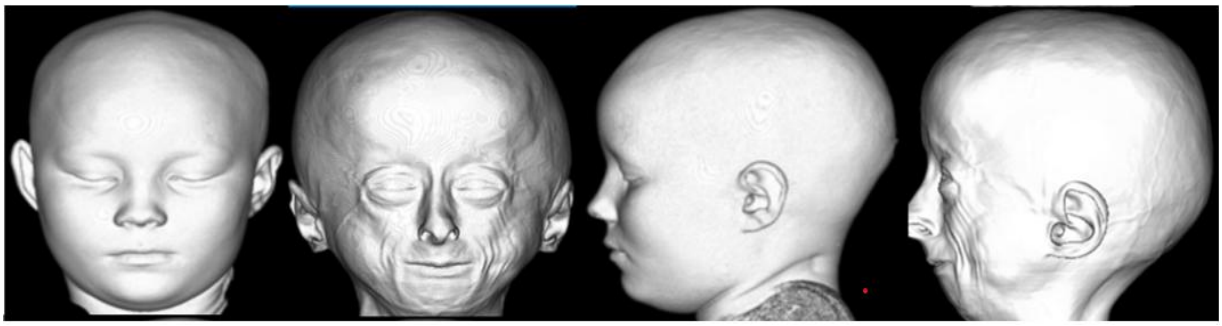

Figure 13.1. Front and side face difference between the normal and HGPS.

Individual facial traits consist of an unusually large head relative to the face, a narrow nasal bridge, a slender nasal tip, thin lip vermilion, a small mouth, retrognathia and micrognathia, delayed primary tooth loss, and incomplete secondary tooth emergence[4]. Many patients experience advancing cardiovascular disease and atherosclerosis, typically linked with older age, ultimately causing strokes or heart attacks, leading to death in their early teenage years. The typical life span of a child with Progeria is approximately 14 years, although some have passed away sooner or even reached their 20s. Progeria, a type of laminopathy, results from a mutation in lamin A encoded by the LMNA gene. In the absence of lonafarnib treatment, mortality (range: 6-20 years) occurs at an average age of 14.5 years[5].

CAUSES[6]:

Mutation in the LMNA gene (c.1824C>T)

↓

A wrong (cryptic) splice site is formed

↓

Abnormal splicing produces faulty protein → Progerin

↓

Progerin remains farnesylated and stays at the nuclear envelope

↓

Weak & misshapen nucleus

↓

Premature ageing of body cells

Mutation predicted by 3DSNP'S Database

Sequence of LMNA protein: 664 Amino acids

>sp|P02545|LMNA_HUMAN Prelamin-A/C OS=Homo sapiens OX=9606 GN=LMNA PE=1 SV=1

METPSQRRATRSGAQASSTPLSPTRITRLQEKEDLQELNDRLAVYIDRVRSLETENAGLR
LRITESEEVVSREVSGIKAAYEAELGDARKTLDSVAKERARLQLELSKVREEFKELKARN
TKKEGDLIAAQARLKDLEALLNSKEAALSTALSEKRTLEGELHDLRGQVAKLEAALGEA
KKQLQDEMLRRVDAENRLQTMKEELDFQKNIYSEELRETKRRHETRLVEIDNGKQREFE
SRLADALQELRAQHEDQVEQYKKELEKTYSAKLDNARQSAERNSNLVGAAHEELQQSR
IRIDSLSAQLSQLQKQLAAKEAKLRDLEDSLARERDTSRRLLAEKEREMAEMRARMQQ
QLDEYQELLDIKLALDMEIHAYRKLLEGEEERLRLSPSPTSQRSRGRASSHSSQTQGGGS
VTKKRKLESTESRSSFSQHARTSGRVAVEEVDEEGKFVRLRNKSNEDQSMGNWQIKRQ
NGDDPLLTYRFPPKFTLKAGQVVTIWAAGAGATHSPPTDLVWKAQNTWGCGNSLRTA
LINSTGEEVAMRKLVRSVTVVEDDEDEDGDDLLHHHHGSHCSSSGDPAEYNLRSRTVL
CGTCGQPADKASASGSGAQVGGPISSGSSASSVTVTRSYRSVGGSGGGSFGDNLVTRSY
LLGNSSPRTQSPQNCSIM

Position of 59 Mutation in the LMNA protein: 25, 33, 35, 39, 43, 45, 50, 63, 65, 112, 133, 140, 150, 189, 190, 196-199, 206, 208, 222, 232, 248, 249, 259, 261, 267, 268, 271, 294, 295, 303, 336, 343, 355, 358, 361, 371, 377, 386, 401, 446, 449, 453, 454, 456, 461, 467, 469, 481, 520, 527, 528, 530, 541, 602, 624, 644.

DIAGNOSIS[5]: History, Physical Examination, Genetic test - a mutation in the LMNA gene

TREATMENT[7]:

At present, there is no lasting remedy for Progeria. The condition results from a modification in the LMNA gene, ensuing in excessive accumulation of the abnormal protein - progerin. Current therapies such as Lonafarnib merely slow disease progression and enhance quality of life, rather than providing a cure.

Investigations continue in these fields: Gene Therapy - Fixing or inhibiting the LMNA mutation. CRISPR-Cas9 technology - Trials conducted to fix the faulty gene. New Drug Development - Merging Lonafarnib with other medications (e.g., everolimus) to diminish progerin toxicity.

TIMELINE FOR RESEARCH ON HGPS [8,10]:

Table 13.1. Timeline For Research on HGPS

2012	Lonafarnib	clinical use
2020 (Nov)	FDA approval of Lonafarnib (Zokinvy)	First authorized medication for HGPS, reduces harmful progerin accumulation.
2023–2024	Preclinical studies (Lonafarnib + Everolimus)	Enhanced models of blood vessel structure and function
2024 (ongoing)	PhaseI/IItrial (Lonafarnib + Everolimus)	Evaluating safety and effectiveness in patients.
2025 (mid-year)	Study on endothelial cells under stress	Identified unusual vascular reaction; gene modification rectified issue.
2025 (ongoing)	Gene editing approaches	Suggested direct amendment of the HGPS mutation within a living organism.
	Stem cell therapy research	Mesenchymal stem cells demonstrate potential in tissue regeneration and alleviating symptoms.

MANAGEMENT[9]:

- Cardiac care - It includes regular checkups, blood pressure control, and statins.
- Bone and joint health – Adequate intake of Vitamin D, calcium, and exercise.
- Nutrition and skincare - Calorie-rich meals, hydrating creams, oral hygiene.
- Infection prevention - Vaccinations, prompt therapy.
- Emotional assistance & challenges - Therapy, engagement in novel treatments

CONCLUSION:

Hutchinson–Gilford Progeria Syndrome results from gene mutations of LMNA leads to the synthesis of defective protein called "progerin," causing organismal and cellular deterioration. HGPS cells exhibit impairment of nuclear functions, encompassing epigenetic changes, dysregulated gene expression, defects in DNA repair, and dysfunction of telomeres. Current research and specific treatments, like lonafarnib, are starting to enhance the quality of life for those impacted. Ongoing investigation into gene editing, stem cell therapies, and molecular treatments may improve approaches for Progeria and reveal tactics to tackle age-related illnesses, resulting in fatalities in their teenage years from heart attacks or strokes caused by advanced atherosclerosis. The expression of progerin rises with age in healthy individuals, indicating its role in physiological aging. Present treatments focus on decreasing progerin toxicity by lowering its levels or directly addressing disrupted cellular processes

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Chapter - 14

A Review of Central Nervous System Autoimmune Disease: Multiple Sclerosis

Vetrivel. A.U

ABSTRACT:

Multiple sclerosis is an enduring autoimmune disorder impacting central nervous system, noticeable by loss of neurons, demyelination, inflammation, and gliosis. This condition represent the diverse array of neurological indicators, including vision problems, localised weakness, bladder and bowel issues, numbness and tingling sensations and cognitive difficulties. There is various illness patterns associated with multiple sclerosis, including primary and secondary progressive, and relapsing-remitting. Timely diagnosis, usually grounded in episodes occurring at different times and locations, enables the swift start of disease-modifying treatment focused on minimising relapses and long-term impairment, essential for correctly identifying multiple sclerosis. Successful management of multiple sclerosis necessitates a collaborative approach and numerous treatment alternatives, such as disease-modifying therapies. This article offers a comprehensive overview of Multiple sclerosis

Keywords: Multiple sclerosis, Causes, Diagnosis, Treatment.

INTRODUCTION:

Multiple sclerosis (MS) is an autoimmune condition marked by the loss of myelin and damage to axons within the central nervous system (CNS). It is twice as prevalent in females and is typically diagnosed between the ages of 20 and 40[1]. In multiple sclerosis, the immune system erroneously assaults the brain and spinal cord. It accomplishes this by harming myelin – the protective layer surrounding the nerves. When myelin is harmed, communications cannot be conveyed properly from the brain and spinal cord to various areas of the body[2].

Around 3 million individuals globally have multiple sclerosis (MS), which is a major reason for neurological disabilities among young adults. MS is marked by injuries in the brain, spinal cord, and optic nerve identified through MRI (magnetic resonance imaging). Lesions in MS are varied, leading to diverse signs based on the specific location[3]. In MS, the immune system is inappropriately triggered, infiltrating in CNS, protective layer surrounding nerve fibres (axons), causing harm to myelin, leading to demyelination. Axons are answerable for carrying

action potentials crucial for relaying data to another neurons across CNS. The myelin sheath controls the velocity of nerve transmission and preserves wellbeing of axon. Consequently, injured myelin not only hinders the transmission of nerve signals, leading to incapacity in MS, but ongoing demyelination sustains axonal injury, a crucial factor is advancement of disease and lasting neurological impairment[4].

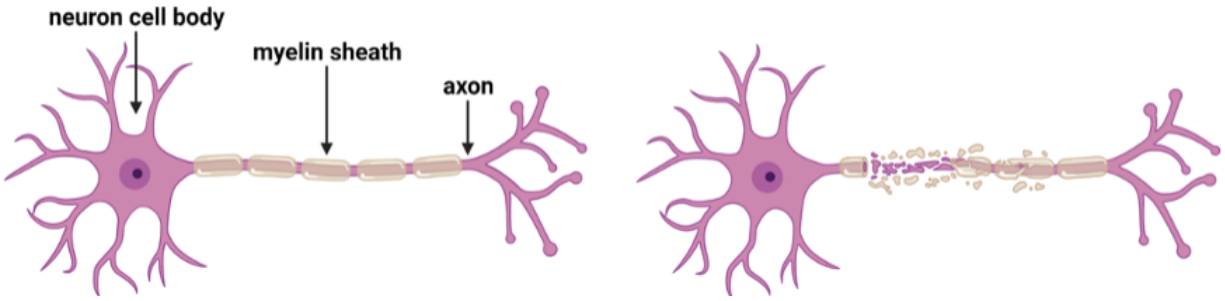

Figure 14.1. Central Nervous System Autoimmune Disease -Multiple Sclerosis

CAUSES[5]:

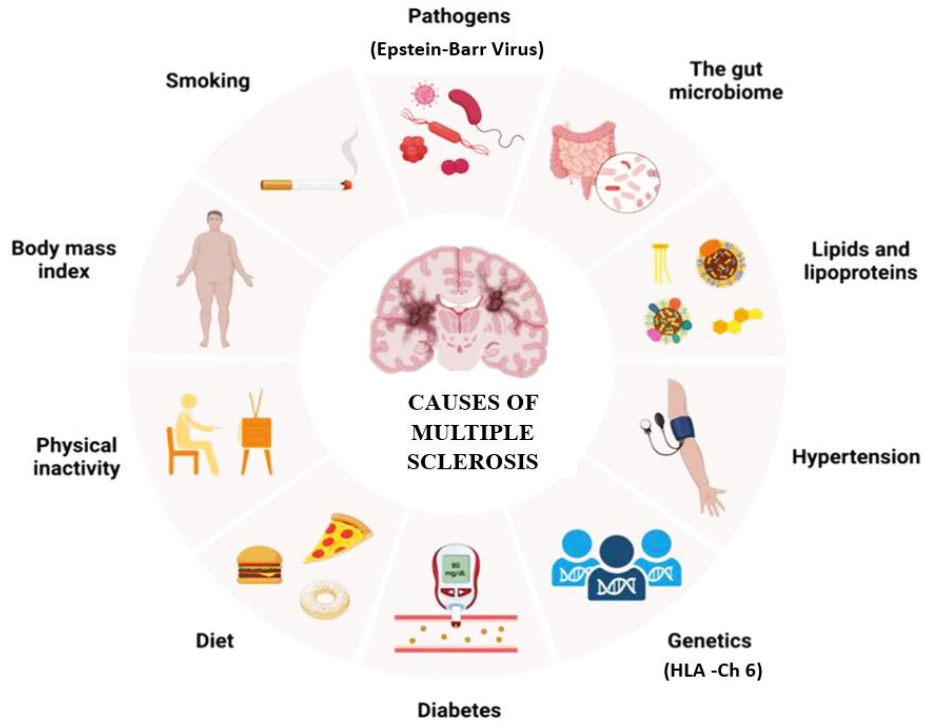

Figure 14.2. Causes of Multiple Sclerosis

DIAGNOSIS[6]:

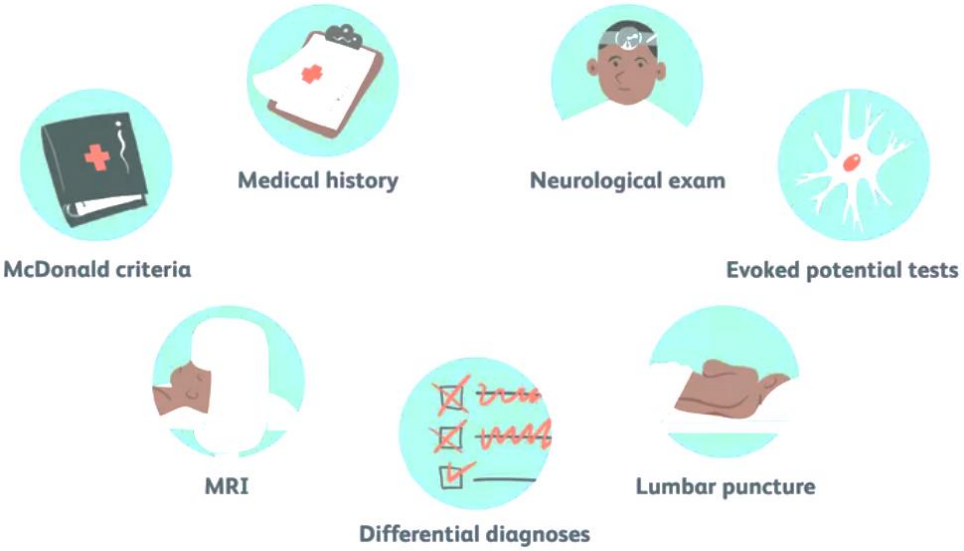

Figure 14.3. Diagnosis of Multiple Sclerosis

McDonald criteria:

The global benchmark for the early and precise diagnosis of multiple sclerosis (MS) by showing MS lesions that are dispersed in time and space in the central nervous system

SIGNS AND SYMPTOMS[7]: The subsequent are initial indicators and symptoms of MS in individuals of all genders - Urinary issues, Digestive troubles, Trouble with focus/concentration, Lightheadedness/poor coordination, Exhaustion, Muscle cramps, Body aches, Speech difficulties, Tingling and numb feelings, Vision difficulties, and Weakness.

TREATMENT[8]:

Table: 14.1 Disease-Modifying Therapies

Common Name	IUPAC Name	Administration	Treatment Duration
Copaxone	Glatiramer acetate	Subcutaneous injection	Extended duration, frequently spanning years, with regular evaluations.
Tysabri	Natalizumab	Intravenous infusion	
Gilenya	Fingolimod	Oral capsule	
Tecfidera	Dimethyl Fumarate		
Zeposia	Ozanimod		
Aubagio	Teriflunomide	Oral tablet	
Mayzent	Siponimod		
Ponvory	Ponesimod		

1. Disease-Modifying Therapies (DMTs): DMTs seek to change the trajectory of MS by adjusting the immune system to diminish inflammation and avert nerve injury. These treatments are mainly employed for managing relapsing types of MS and are given via different methods depending on specific symptoms and phases.

2. Symptomatic Treatments: In addition to DMTs, symptomatic treatments are employed to manage specific MS symptoms:

Table: 14.2. Symptomatic Treatments

Common Name	Drug Name	Treatment
Corticosteroids	Methylprednisolone	Employed to reduce inflammation during relapses.
Muscle Relaxants	Baclofen, Tizanidine	Tackle spasticity.
Antidepressants	Sertraline, Fluoxetine	Handle depression and emotional fluctuations.
Pain Relievers	Gabapentin, Pregabalin	Relieve neuropathic discomfort.
Bladder Medications	Oxybutynin	Address urinary symptoms.

RECENT ADVANCES IN RESEARCH[9,10]:

Table: 14.3. Recent Advances in Research (Multiple Sclerosis)

THERAPY	METHOD	MECHANISM	RESULT
Tolebrutinib	New Drug for Progressive MS	Target the particular immune cells engaged to provide a focused treatment strategy	Potential to reduce the advancement of progressive MS by 31%
Fenebrutinib	New Drug for Relapsing MS	Focusing on specific immune cells involved in MS,	95% of individuals with relapsing MS who received the drug experienced no relapses over 48 weeks.
Combined Therapy	Vitamin D_3 and Dimethyl Fumarate	Improve the efficacy of MS treatment.	Assist in regulating the immune system, decreasing relapse occurrences, and minimising lesion development.
Intranasal Foralumab	A nasal spray	Aims at particular immune cells related to MS.	Efficiently adjust the immune system and lessen neuroinflammation while maintaining a positive safety profile.
Ocrelizumab	New Injection Form	Enhances the convenience of treatment for patients.	Patients will get the medication through a fast 10-minute injection rather than a 4-hour infusion

CONCLUSIONS:

In summary, Multiple Sclerosis (MS) is a complicated, incurable autoimmune condition with a highly unpredictable progression, yet recent advancements in treatment have significantly enhanced patient prognosis. Although there is no cure, effective disease-modifying therapies (DMTs) can greatly control the inflammatory phase, particularly in relapsing-remitting MS (RRMS), enabling many patients to lead complete, productive lives with nearly normal life spans. Nonetheless, a considerable unmet requirement persists for halting the neurodegenerative progression observed in the later phases of the illness

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Chapter - 15

Congenital Sensory Defects (Birth Anomalies): A Literature Review

Sahana. R

ABSTRACT:

Congenital sensory impairments can be described as structural or functional irregularities of the senses (hearing, vision, touch, taste, smell, or balance) that arise during prenatal development. Congenital sensory impairments represent a major health concern affecting no fewer than five million births each year globally. Approximately 3 to 4 % of infants are born with a variety of birth defects. Birth defects can range from very mild to quite severe. Certain birth defects can be fatal, meaning a baby might only survive for a few months. These conditions arise during prenatal development and can be detected before or at the time of birth, or at a later stage in life. Nonetheless, the actual count of cases might be significantly greater as statistics frequently exclude terminated pregnancies and stillbirths. This article explored the symptoms of various types of congenital sensory defects, including their diagnostic techniques and treatment options.

Keywords: Congenital sensory defects, Symptoms, Diagnosis, Treatment.

INTRODUCTION:

Congenital sensory impairments can lead to enduring disabilities, significantly affecting individuals, families, healthcare systems, and communities. Approximately 240,000 infants globally perish within 28 days of birth each year as a result of congenital anomalies[1]. Congenital sensory disorders result in an additional 170,000 fatalities among children aged 1 month to 5 years. Nine out of ten children born with a severe congenital condition are located in low- and middle-income nations[2]. With the reduction of neonatal and under-5 mortality rates, congenital disorders account for an increasing share of the reasons for neonatal and under-5 fatalities. Despite congenital sensory disorders potentially arising from various genetic, infectious, nutritional, or environmental influences, pinpointing the precise causes is frequently challenging. Certain congenital sensory impairments can be avoided[3]. Vaccination, sufficient consumption of folic acid or iodine via food fortification or supplements, and proper care before and during pregnancy are examples of preventive measures[4].

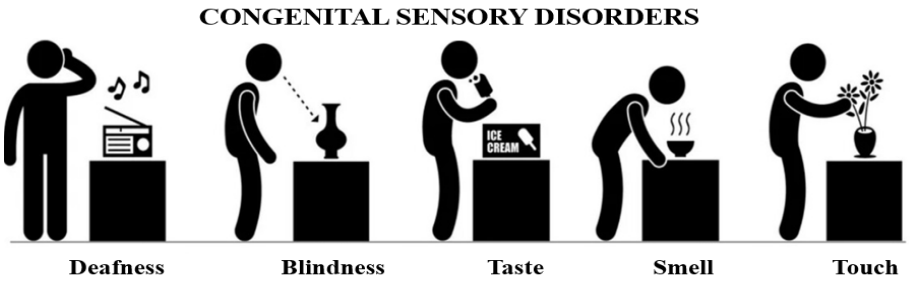

Figure 15.1. Congenital Sensory Disorders

In recent decades, numerous prevalent historical factors leading to congenital sensory impairments have been eliminated or reduced due to economic growth and specific initiatives addressing disease and maternal health, among others. Consequently, at a certain moment within the past few decades, and possibly for the first time in human history, inherited conditions have emerged as the leading cause of congenital sensory deficits[5]. Congenital sensory impairments represent a significant contributor to the worldwide disease burden, with low- and middle-income nations being disproportionately impacted. The symptoms and issues resulting from congenital anomalies differ, and so do the treatments. Options include medications, therapies, surgeries, and assistive devices[6].

SYMPTOMS[7-9]:

Table. 15.1. Symptoms of Congenital Sensory Disorders

Hearing Impairment (Deafness)	Visual Impairment (Blindness)	Tactile Sensory Impairment (Touch)	Olfactory Impairment (Smell)	Gustatory Impairment (Taste)
• Child is born with a partial or complete inability to hear sounds. • Lack of Response to Sound. • Delayed Speech Development. • Failure to Respond to Their Name. • Difficulty with Language and Understanding.	• Birth can range from partial to complete blindness. • Lack of Eye Contact or Tracking Movements. • Unusual Eye Movements. • Reduced Interest in Visual Stimuli. • Lack of Visual Discrimination.	• Congenital Insensitivity to Pain (CIP) or certain forms of peripheral neuropathy. • Lack of Pain Response. • Delayed Development of Fine Motor Skills. • Lack of Sensory Feedback. • Self-Injurious Behaviour.	• Developmental abnormalities affecting the olfactory system (smell). • Failure to React to Odors. • Lack of Smell-Based Exploration. • Difficulty with Taste Preferences.	• Affecting the gustatory receptors or neural pathways involved in taste perception. • Lack of Interest in Eating or Drinking. • Failure to Respond to Different Tastes. • Feeding Difficulties.

DIAGNOSTIC METHOD[8]:

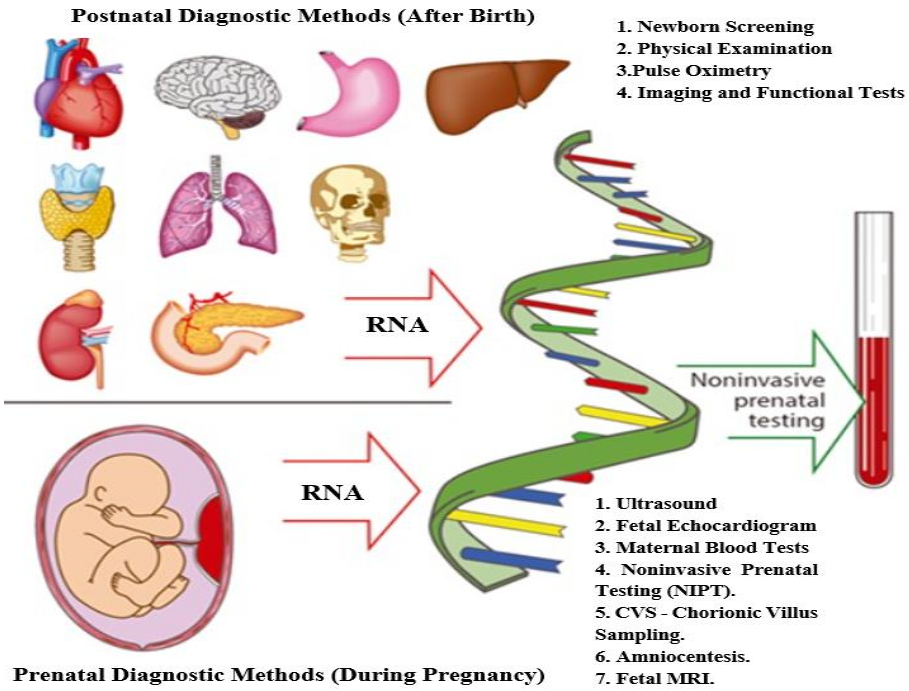

Figure 15.2. Diagnostic Method

CAUSES[6]: Genetic anomalies, Infections in pregnancy, lifestyle decisions and habits, Exposure to certain medications and toxins, Pre-existing health issues such as diabetes

TREATMENT[9]: The majority of congenital deficit loss cannot be addressed with medications, though certain instances may improve with the following:

1. Medications:
   - Corticosteroids – Administered when hearing loss results from inflammation or autoimmune disorders.
   - Antiviral medications – In congenital CMV (Cytomegalovirus) infection, which may lead to sensorineural hearing impairment.
   - Acetazolamide – Administered for Leber's Hereditary Optic Neuropathy (LHON) or optic nerve head drusen to lower intracranial pressure.
   - Idebenone – Uniquely authorized for LHON, a mitochondrial condition leading to abrupt loss of vision.
   - Antipsychotic medications – Occasionally utilized in autism spectrum disorder (ASD) to address irritability or aggression.

2. Surgery:

Applicable for structural problems like congenital anomalies (heart - closing gaps, fixing valves), cleft lip/palate, and spina bifida.

3. Assistive Devices: Orthopaedic supports, auditory aids, or cochlear devices might be necessary.

4. Therapies: Physical therapy, speech therapy, and occupational therapy enhance functional skills and growth

RECENT RESEARCH[9,10]:

Recent studies have resulted in notable progress in treating congenital sensory impairments, especially concerning hearing and vision.

- Gene Therapy for OTOF-Related Deafness:

  A landmark research published in The Lancet showed the success of gene therapy in regaining hearing in children with congenital deafness due to mutations in the OTOF gene.

- Dual-AAV Approach for Large Genes:

  Harvard Medical School researchers have created a dual-AAV gene therapy to tackle the issue of large genes such as OTOF, which surpass the packaging limits of a single AAV vector.

- Gene Therapy for Usher Syndrome Type 1F:

  This an uncommon condition that leads to ear damage and gradual vision impairment. In animal studies, this treatment revived hearing and demonstrated promise for enhancing vision, providing optimism for those with this combined sensory loss.

- Gene Therapy for Norrie Disease:

  Norrie disease is an uncommon hereditary condition that leads to vision loss and advancing hearing impairment. Scientists at University College London have created a gene therapy that stops the death of hair cells in the cochlea, which is the area of the inner ear that enables hearing.

- Gene Therapy for Leber Congenital Amaurosis (LCA):

The treatment included delivering a normal version of the AIPL1 gene into the retina. Gene therapy may serve as a revolutionary method for addressing severe blindness in children.

CONCLUSION:

The primary aspect of managing congenital sensory disorders is prompt diagnosis and timely intervention. Assessment of congenital sensory disorders begins with screenings of newborn sense organs for Acute Bacterial Rhinosinusitis (ABRs) and involves a comprehensive history and physical examination, diagnostic imaging, electrocardiography, genetic testing and consultation, along with an ophthalmologic examination. The main approach to treatment includes sensory implantation and amplification, along with language therapy in a suitable educational setting. At the national level, they can aid in the proper formulation of programs for the prevention and management of congenital sensory disorders to enhance treatment in prenatal care

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Chapter - 16

An Assessment on Lung Cancer

Mahaa Shree. P.K, Nithyasai. K, Aishvaryaa. N, Sanjana. A

ABSTRACT:

Lung cancer ranks among the most prevalent malignant tumors in many nations and is the primary cause of cancer-related fatalities globally. In spite of ongoing advancements in diagnosis and treatment, the survival rates for individuals diagnosed with lung cancer are still inadequate. Lung cancer is the main trigger of cancer-related deaths in both genders, as stated in the WHO report with 2.09 million cases worldwide. It is the primary reason for cancer mortality in men and the 2nd most common source of cancer death in women globally. Lung cancer treatment can include a mix of surgery, chemotherapy, targeted therapy, immunotherapy, and radiation therapy. This review aims to summarize the phases, categories, and therapies of lung cancer

Keyword: Lung Cancer, Stages, Types, Treatment.

INTRODUCTION:

Cancers are defined by their uncontrolled proliferation and dissemination of cells to various areas in the body. Lung cancer is among the most prevalent cancers, and a significant number of individuals succumb to this illness annually[1]. The illness is frequently identified at a later phase, yet even in earlier phases, patients with lung cancer experience poorer results than those with different types of cancer. Even without metastasizing to another organ, the rate of survival for lung cancer in stage I is <70%[2].

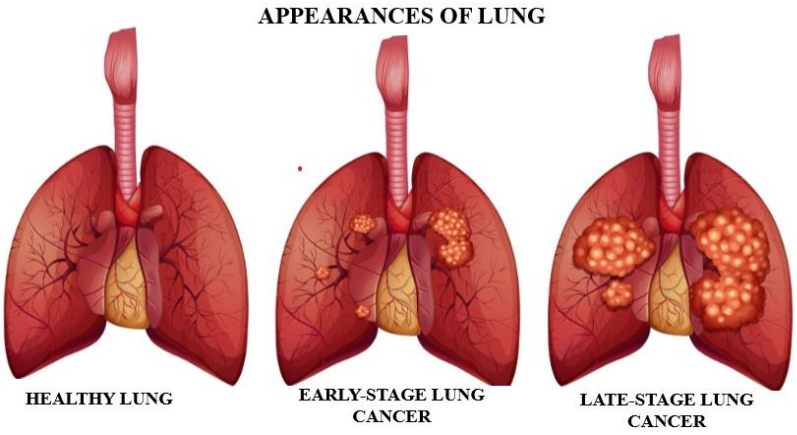

Figure 16.1. Appearance of Lungs

Lung cancer arises from an irregularity development of the cell. Typically, the human body regulates a set of regulator systems for cell growth, ensuring that cells only divide to generate new ones when there is a need for additional cells[3]. Interference with this regulatory system leads to unregulated cell division and growth, ultimately creating a mass referred toas a tumor. While it can arise in any area of the lung, lung cancers (90-95%) originate from bronchioles, bronchi and epithelial cells[4,5]. It may occasionally develop from other supportive tissues in the lungs, such as blood vessels. These cancers develop from a multistage process that encompasses numerous epigenetic and genetic alterations, including the impairment of various essential cell-cycle genes. The changes can build up in bronchial epithelium resulting in clonal cell growth[6].

MECHANISM OF LUNG CANCER[7,8]:

Lung cancer is caused by variation in the DNA of lung cells. DNA comprises the guidelines that inform a cell how to function. In normal cells, DNA provides directions for growth and replication at a predetermined pace. The directives instruct the cells to perish at a designated time. In cancerous cells, the modifications in DNA deliver various instructions for rapid produce a larger number of cells. Cancer cells can continue to survive while healthy cells would succumb. This results in an excess of cells. Cancer cells can generate a lump known as a tumor which has the potential to expand, infiltrating and damaging normal body tissue. Eventually, cancer cells may disperse and detach to different parts of the body. Once the cancer advances, it's called as metastatic.

STAGES OF CANCER[9]:

The initial site of spread for the majority of lung cancer patients is the lymph nodes. Evaluating the potential for lymph node involvement before surgery is crucial for planning additional treatment. Lymph nodes will be excised during the operation for analysis via light microscopy for evaluation. If tumor cell regions are present in the lymph nodes, the patient receives extra chemotherapy. In many instances, the dissemination of cancer to lymph nodes goes undetected because of the ineffectiveness of clinical techniques, resulting in a low survival rate for patients, even post-diagnosis. Greater sophistication and sensitivity in methods are necessary to assist with this

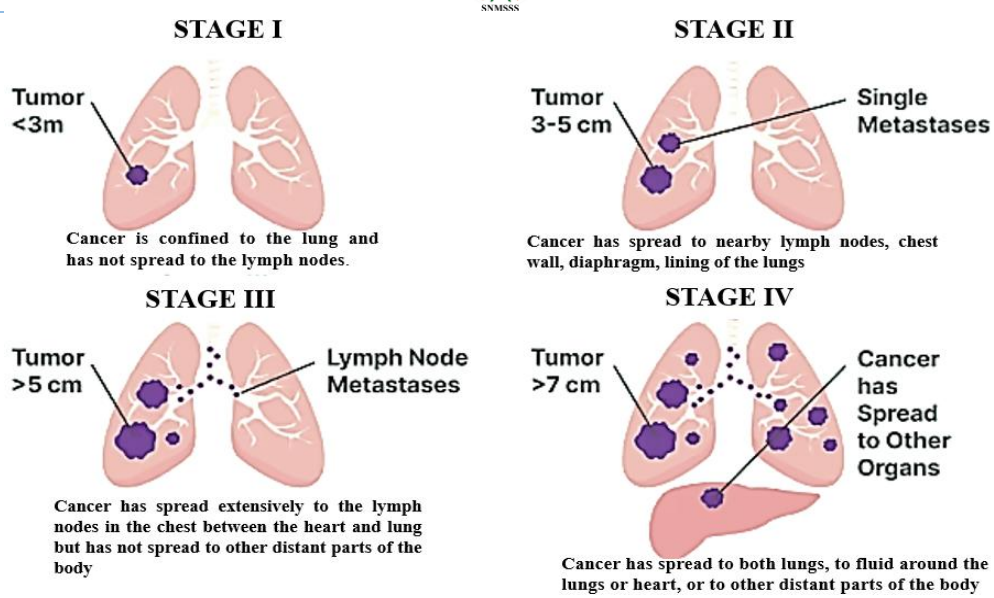

Figure 16.2. Stages of Cancer

CAUSES OF LUNGS CANCER[10]:

Comprehending the elements that lead to lung cancer aids in prevention and minimizing risk.

1. Tobacco and Smoking: Cigarette use is the primary factor behind lung cancer, accounting for most instances. The harmful substances in tobacco harm lung tissue, greatly elevating the likelihood of cancer. Even passive smoking can raise the risk of lung cancer, particularly if exposure is extended.

2. Environmental & Occupational Exposure: Contact with hazardous materials like asbestos and specific industrial chemicals raises the risk of lung cancer. Although radon exposure is a major factor in certain areas of the globe, it is not as prevalent in India. Air pollution could also increase risk, although its causal relationship is still under investigation.

3. Lifestyle and Additional Risk Factors: Existing respiratory issues, like chronic obstructive pulmonary disease (COPD), combined with unhealthy eating habits or lack of exercise, can heighten susceptibility. Sustaining a healthy lifestyle can lower the risk, yet it does not completely remove the chance of getting lung cancer

TYPES OF LUNGS CANCER[11]:

80% of patients having NSCLC type

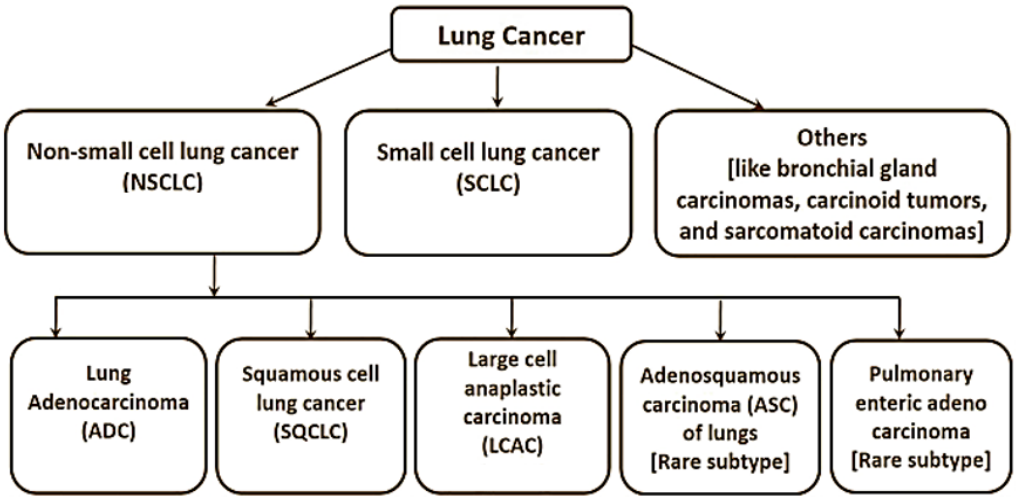

Figure 16.3. Types of Cancer

DIAGNOSIS OF LUNG CANCER[12]:

1. Standard evaluation: Medical history and physical examination, Chest radiograph, Computed Tomography (CT) Scan.

2. Strong suspicion of lung cancer:

- Bronchoscopy: The physician places a slender, illuminated tube (a bronchoscope) through the mouth or nose into the lung. The physician might collect a cell sample using a needle, brush, or another instrument. The doctor might also rinse the area with water to gather cells in the liquid.
- Thoracocentesis: When fluid accumulates around the lungs (pleural effusion), the physician can obtain some fluid using a needle inserted through the skin.
- CT-guided biopsy: The physician employs a slender needle to extract tissue or fluid from the lung with the assistance of a CT scan.
- Open biopsy: When it is challenging to acquire the tumour tissue, a direct biopsy of the lung tumour or lymph nodes may require an incision in the chest wall.

SIGN AND SYMPTOMS OF LUNG CANCER[10]:

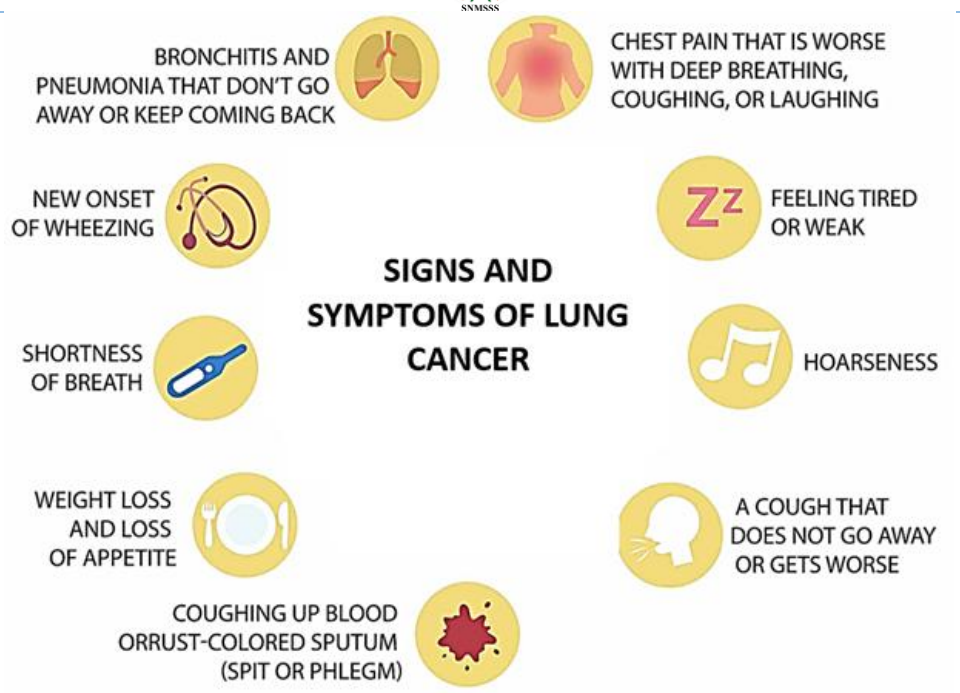

Figure 16.4. Sign and Symptoms of Lung Cancer

TREATMENT OF LUNG CANCER[13]:

Depending on the lung cancer stage, treatment options can differ to enhance quality of life.

- Lung Cancer Screening: To identify cancers early for timely treatment at an initial stage.
- Surgery: It includes the excision of tumor-containing tissues and adjacent lymph nodes. This can be achieved through wedge resection (removal of a segment of lung), lobectomy (removal of a complete lobe of the lung), or pneumonectomy (removal of the whole lung).
- Chemotherapy: Chemotherapy utilises anticancer medications to destroy cancer cells. This is typically administered as an infusion. The medications enter the circulatory system and can influence cancer cells throughout the entire body.
- Radiation therapy: It services high-energy beams to eradicate cancer cells. It impacts cells solely in the targeted region. For individuals with advanced illness, it can be utilised before or following surgery, and is frequently paired with chemotherapy.
- Targeted Treatment: Targeted treatment concentrates on particular anomalies found in cancer cells. Targeted drug treatments can lead to the death of cancer cells by inhibiting these abnormalities.

  Drug used: Osimertinib, Crizotinib, Capmatinib.

- Immunotherapy: A kind of cancer therapy that strengthens the body's defenses against malignant cells. Cancer cells possess the capability to 'disguise' themselves, making it difficult for our body's immune system to identify and eliminate these 'rogue cells.'

Drug used: Atezolizumab, Durvalumab, Nivolumab, Pembrolizumab.

CONCLUSION:

In this overview, we have succinctly described lung cancer. It is the top reason for cancer fatalities and the 2nd most frequently detected cancer in both genders. Lung cancers usually begin in the cells that areas of the lung and line the bronchi, including the alveoli or bronchioles. Lung cancer can be addressed in various ways, based on the specific stage and type of the disease. Individuals with non-small cell lung cancer may serve through chemotherapy, targeted therapy, surgery, radiation therapy or a mix of these approaches. Individuals diagnosed with small-cell lung cancer typically receive radiation therapy and chemotherapy as part of their treatment

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Chapter - 17

Review on a Bombay Blood (Rare Blood) Group

Sanjai. K

ABSTRACT:

The Bombay blood group is a unique form of blood type that is quite different from the ABO system and was initially identified in India around five decades ago. It is marked by a lack of typical ABH antigens and has been consistent with serum antibodies. This blood type is suspected when reagent O cells demonstrate agglutination in reverse or back typing or during antibody screening. This blood type pertains to people who do not have the H antigen, resulting in the presence of anti-H antibodies along with anti-A and anti-B antibodies. Members of this group can exclusively accept blood from other Bombay donors due to the presence of strong anti-H antibodies that trigger serious transfusion reactions to standard blood types. It is frequently mistaken for Group O since standard ABO testing (forward grouping) fails to identify A/B or antigens, resulting in no reaction. Administering non-Bombay blood (such as Type O) causes serious haemolytic transfusion reactions.

Keywords: Bombay Blood group, H antigen, Biochemistry, Genetics.

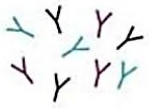
INTRODUCTION:

The Bombay blood group, known as hh, lacks the expression of antigen H. This indicates that the red blood cells of the hh blood group do not contain antigen H. The hh blood group is frequently mistaken for the O blood group[1]. The distinction lies in the fact that the O group contains Antigen H, whereas the hh group lacks it. The unique Bombay blood group was initially identified in Mumbai (formerly Bombay) in 1952 by Dr. Y M Bhende. Worldwide, the hh blood type occurs in one out of four million individuals[2].

There has been a recent increase in the demand for a rare blood type known as the Bombay blood group. In the ABO blood group system, blood types are divided into four main groups: A, B, AB, and O. Each red blood cell features antigens on its surface that help establish its group. For example, the AB blood type contains both A and B antigens. A possesses A antigens; B contains B antigens. In blood type O, A and B antigens are absent. The A, B, and O blood types were initially discovered by Austrian immunologist Karl Landsteiner in 1901[3].

The Bombay blood group is found more frequently in South Asia than in other regions due to inbreeding and marriages within tight-knit communities. In India, one individual out of 7,600 to 10,000 is born with this variety. Because hh blood type is so uncommon, patients encounter difficulties during blood transfusions, frequently resulting in death due to the lack of hh blood. People with the Bombay blood group can receive transfusions exclusively from others with the Bombay hh phenotype, which is extremely uncommon[4]. Rejection can happen if they obtain blood from A, B, AB, or O blood types. Conversely, the blood type can provide blood to the ABO blood groups. This blood type is typically not kept in blood banks, mainly due to its rarity and the fact that blood has a shelf life of 35-42 days. Since this condition is extremely uncommon, anyone with this blood type who requires an immediate blood transfusion will likely face difficulties obtaining it, as no blood bank is expected to have it available[5].

Table 17.1. Types of Blood Group

BLOOD GROUPS					
	Type A	**Type B**	**Type AB**	**Type O**	**Type Bombay O**
Antigen (on RBC)	Antigen A 	Antigen B 	Antigen A + B 	Antigen H 	No Antigen 
Antibody (in plasma)	Anti-B Antibody 	Anti-A Antibody 	Neither Antibody	Anti-A & Anti-B 	Anti-A, Anti-B and Anti-H 
Cannot donate	O, B, Bombay O	O, A, Bombay O	O, A, B, Bombay O	Bombay O	
Can donate	A, AB	B, AB	AB	O, A, B, AB	O, A, B, AB Bombay O
Can receive	A, O	B, O	AB, A, B, O	O	Bombay O

There has been a recent increase in the demand for a rare blood type known as the Bombay blood group. In the ABO blood group system, blood types are divided into four main groups: A, B, AB, and O. Each red blood cell features antigens on its surface that help establish its group. For example, the AB blood type contains both A and B antigens. A possesses A

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GENETICS[6]:

The Bombay phenotype is found in individuals who possess two recessive alleles of the H gene (meaning their genotype is hh). These people do not generate the H carbohydrate, which is essential for the formation of A and B antigens, indicating that someone can have alleles for one or both of the A and B variants without being able to manifest them. Since both parents need to possess this recessive allele to pass on this blood type to their offspring, the condition predominantly arises in isolated small communities where it's likely for both parents of a child to either be of Bombay type or have heterozygosity for the h allele, thus carrying the Bombay trait as recessive. Other instances might involve aristocratic families, which are inbred due to tradition rather than regional genetic diversity.

Table 17.2. ABO groups vs Bombay blood group

ABO groups	**Bombay blood group (hh)**
Influenced by variants of the ABO gene (A, B, or O alleles).	Even if an individual possesses A or B alleles, they are unable to express them.
ABO group is presumed that H antigen (through FUT1) is present.	FUT1 gene mutation inhibits the production of H antigen. Genetically A/B/AB may show a phenotype of O unless an anti-H test is performed.

BIOCHEMISTRY[7]:

The biosynthesis of H, A, and B antigens entails enzymes that move monosaccharides, leading to oligosaccharide chains linked to lipids and proteins in red blood cells. The specificity of the H antigen is dictated by the arrangement of oligosaccharides, with the essential component being the terminal disaccharide of fucose-galactose.

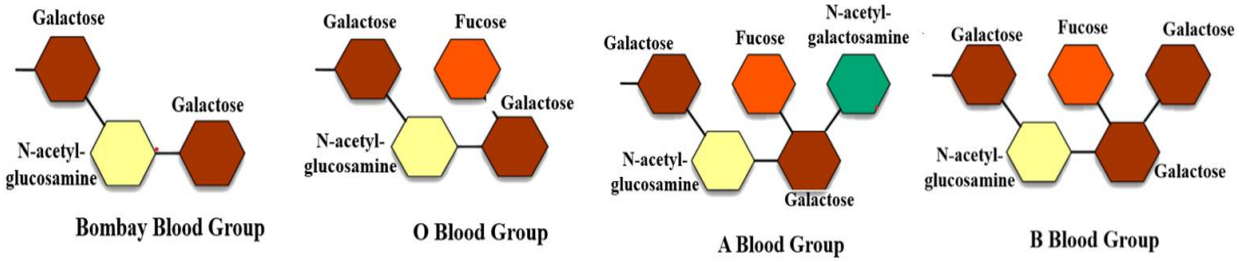

Figure 17.1. Biochemical struture of Blood Groups

Every human red blood cell, with very few exceptions (such as the Bombay blood group), possesses the red cell H antigen. It is found in the highest concentration on O type cells and the lowest on AB type cells. H constituent is generated biochemically through the attachment of fucose to glycoproteins on the outward, with the progression facilitated by Fucosyl transferase. When N-Acetyl galactosamine attaches to the H constituent, it creates A blood conversely, when galactose attaches, it results in B group. The lack of any essential element for H leads to the formation of O blood. The phenotype of the Bombay is defined by a lack or mutations of Fucosyl transferase

TRANSFUSION CHALLENGES[8]:

People with the Bombay phenotype do not have H antigen, a precursor to the A & B antigens within the ABO group system. Consequently, they are unable to display A or B antigens irrespective of their genotype. Their immune systems create antibodies against the H antigen, which means they cannot accept RBC from ABO group donor, including group O. Only blood from another Bombay phenotype person is suitable. Routine blood typing might wrongly classify them as group O unless specialized anti-H testing is conducted, which is essential to prevent serious transfusion reactions.

Compatibility of hh (Bombay) Blood Group[9]:

As a Beneficiary

- Can accept - Only from hh (Bombay) contributors.
- Unable to receive - From O, A, B, or AB donors (including O negative), as all possess the H antigen, which would provoke a severe immune response.

As a Contributor:

- Can contribute to: Other hh (Bombay) persons exclusively.
- Unable to donate to: O, A, B, or AB groups, as their immune systems anticipate the H antigen, which hh blood cells do not possess.

Table 17.3.Blood Antigens and Antibodies

Blood Groups (Antigen and Antibodies)		
Blood Group	**Antigens**	**Antibodies**
A	A,H	B
B	B,H	A
AB	A,B,H	-
O	H	A,B
Bombay blood group	-	A,B,H

Immunity & Differences: Bombay vs ABO Groups[10]:

ABO blood types:

Incompatible reactions happen when A or B antigens that do not match are transfused.

Example:

An individual with type A blood receiving type B blood → anti-B triggers hemolysis.

Bombay (hh):

Transfusion with any ABO blood causes anti-H antibodies to target donor red blood cells. This leads to a serious, quick hemolytic transfusion reaction, even with O blood (which contains H antigen). Consequently, their immunity is more limited than that of ABO groups

CONCLUSION:

The hh (Bombay) blood type is among the rarest and most clinically important blood phenotypes recognized by science. In contrast to the typical ABO blood groups, Bombay individuals have no H antigen, which prevents the expression of A or B antigens and leads to high levels of anti-H antibodies in their plasma. This specific immunological profile leads to

severe transfusion incompatibility, as they cannot accept blood from any ABO donor-even group O-and can only receive from other hh individuals.

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Chapter - 18

A Review on Rare Blood Disorder – Methemoglobinemia (MetHb)

Thejas. D

ABSTRACT:

Methemoglobinemia is a serious condition where a person cannot efficiently deliver oxygen to body tissues, leading to the creation of an excessive amount of methemoglobin (MetHb). This is primarily due to MetHb being the modified form of haemoglobin (Hb), where the ferrous iron in heme has been oxidized to the ferric form. Methemoglobinemia is associated with numerous medications, chemicals, and agents including local anaesthetics, industrial compounds, and pesticides. Numerous compounds lack composition details and may lead to significant methemoglobinemia. Prompt use of the antidote, methylene blue, when necessary, significantly lowers methemoglobin levels to acceptable ranges

Keywords: Haemoglobin, methemoglobinemiab(MetHb), Ferric ions.

INTRODUCTION:

Methemoglobinemia (MetHb) is an exceptionally uncommon blood condition that influences the ability of red blood cells to transport oxygen to tissues and cells. MetHb can be dangerous, particularly for infants suffering from a severe form of the disorder or individuals who consume recreational substances[1]. Typically, healthcare professionals manage methemoglobinemia with a drug that lowers methemoglobin levels and alleviates symptoms. This condition can be inherited, but MetHb typically occurs due to the use of specific medications, recreational drugs, or exposure to certain chemicals[2].

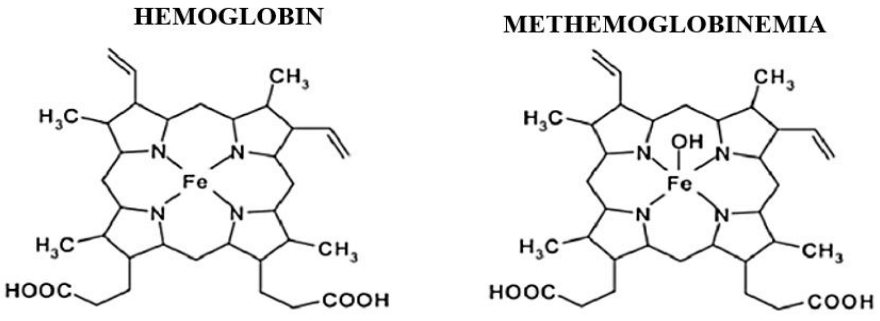

Figure 18.1. Structure of Haemoglobin and Methemoglobinemia

Typically, red blood cells (RBC) transport oxygen across the body, and they depend on the protein haemoglobin for this oxygen transport. In Methemoglobinemia, an excess of methemoglobin is produced, which is a type of haemoglobin, the molecule found in red blood cells that transports oxygen. Nevertheless, methemoglobin is unable to transport oxygen efficiently, resulting in decreased oxygen supply to tissues[3,4].

CAUSES:

Mutations in the HBB gene result in beta-globin type methemoglobinemia. HBB gene delivers the blueprint for producing a protein known as beta-globin. Beta-globin is one of the 4 subunits that constitute haemoglobin. In adults, haemoglobin typically consists of 2 beta and alpha globin subunits. Every protein subunit is attached to a heme, which is an iron-containing elements each heme has an iron atom at its core that can attach to a single O_2 molecule.

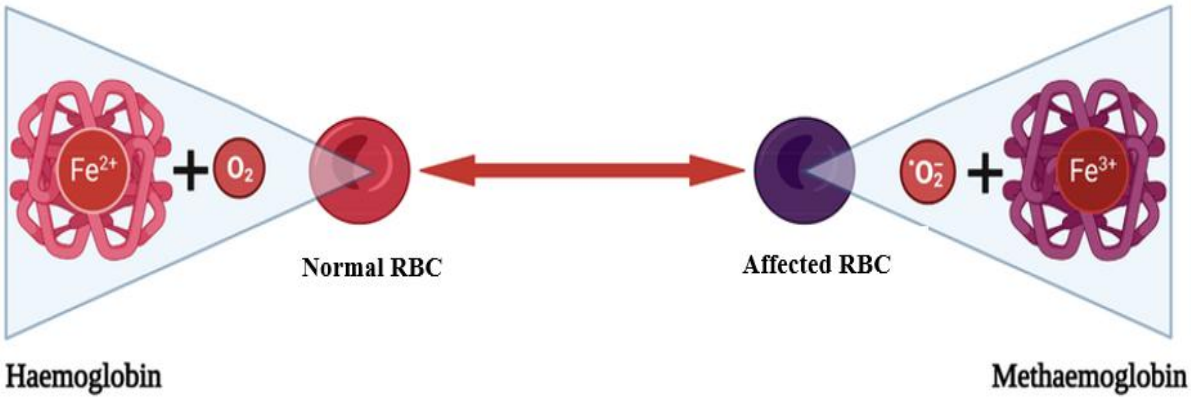

Figure 18.2. Binding of Iron with Haemoglobin and Methemoglobinemia

The iron in the heme molecule must be ferrous iron (Fe2+) in order for hemoglobin to bind to oxygen. Heme can change into ferric iron (Fe3+), a distinct kind of iron that cannot bind oxygen. Ferric iron-containing hemoglobin is called methemoglobin. Methemoglobinemia is caused by mutations in the HBB gene that change the structure of beta-globin and make it easier for heme iron to change from ferrous to ferric. Because ferric iron cannot bind oxygen, cyanosis and dark blood are the results[5].

TYPES[06]:

Table: 18.1. Types of Methemoglobin

Types	Acquired Methemoglobin	Inherited Methemoglobin	
		Type 1	**Type 2**
Alternative names	Blue baby syndrome	Erythrocyte reductase deficiency	Generalized reductase deficiency, Hemoglobin M disease
Causes	Exposed to certain chemicals and medicines (benzocaine, Nitrobenzene, antibiotics).	When RBCs lack the enzyme	When the enzyme doesn't work in the body

SIGN[07]:

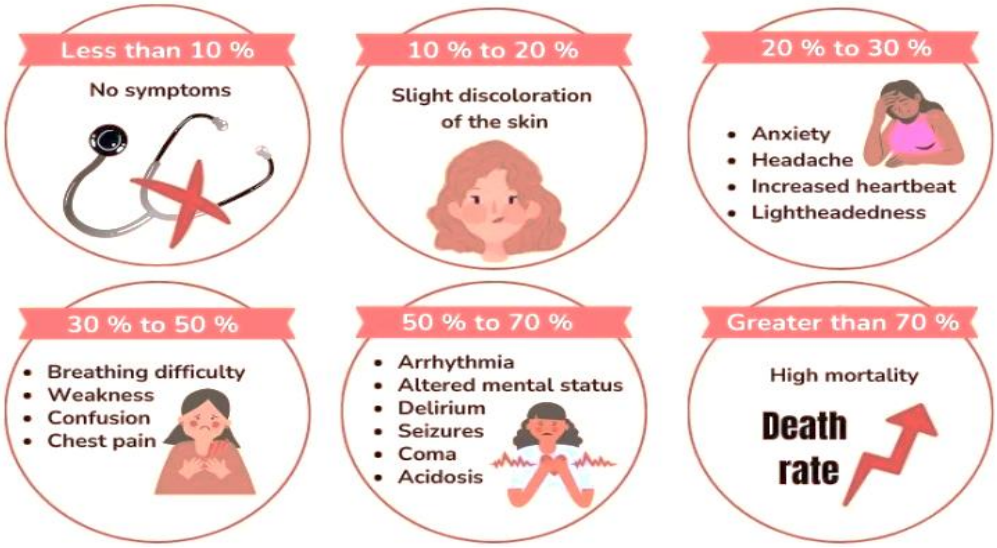

Figure 18.3. Sign of Methemoglobinemia

DIAGNOSIS[08]:

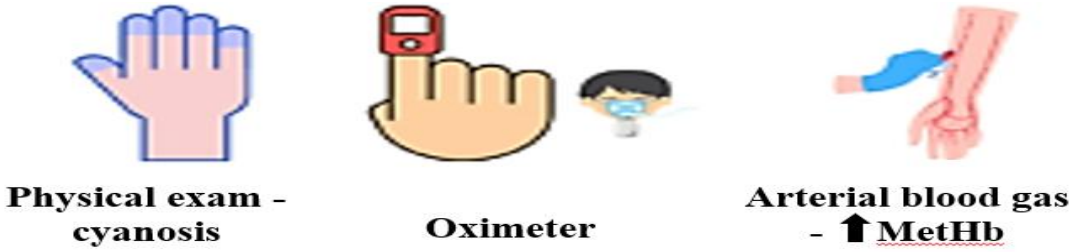

Figure 18.4. Diagnosis of Methemoglobinemia

TREATMENT[09]:

Table 18.2. Treatment of Methemoglobinemia

Drugs	Methylene Blue:	Ascorbic Acid (Vitamin C)
Adults	1–2 mg/kg IV over 5 minutes	300–1000 mg IV or PO daily
children:	1–2 mg/kg IV over 5 minutes	weight- based dosing
Duration of action	Within 30–60 minutes	May take several hours to days for full effect
Max dose	Do not exceed 7 mg/kg,	-

Exchange Transfusion:

- Particularly when Methylene blue fails to work.
- Not recommended (e.g. G6PD deficiency)
- Methemoglobin levels exceeding 70% or persistent symptoms despite therapy.

PREVENTION[10]:

Gene therapy is recommended for duos who have a family background of MetHb and are thinking about taking kids. Infants aged 6 months or less have a higher risk of developing methemoglobinemia. Consequently, homemade baby food purees prepared from vegetables rich in natural nitrates, like carrots, spinach, or beetroots, should be avoided.

AWARENESS ACTIVITY[10]:

- Comprehending the situation
- Identifying signs
- Recognizing risk factors
- Avoiding exposures
- Requesting immediate medical care.

CONCLUSION:

Methemoglobinemia is not regarded as anemia. It is a type of blood disorder in which the Hb is unable to function correctly. Methemoglobinemia may be either hereditary or developed. Chronic congenital methemoglobinemia lacks a pharmacological treatment, whereas acquired methemoglobinemia requires immediate intervention with methylene blue. Nonetheless, those with HbM deficiency do not respond well to methylene blue treatment and might experience the opposite effect. Therefore, individuals with methemoglobinemia need to be evaluated for any additional issues that might contraindicate methylene blue treatment. Individuals prone to methemoglobinemia should seek advice from their physician before starting any new medications and adhere closely to the prescribed treatment if the medications are necessary.

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Chapter - 19

Alopecia: Understanding Hair Loss and its Impact

Sabarish. S

ABSTRACT:

Alopecia is a condition that results in temporary, non-scarring hair loss while preserving the hair follicle. Alopecia is frequent and distressing issue encountered in primary care and can result from numerous causes. Various forms of hair loss consist of clearly defined patches, widespread hair thinning, and complete hair loss that may impact all areas with hair. The most common form of alopecia is characterized by patchy hair loss on the scalp. Hair loss, or alopecia, impacts a large portion of the population at some point in life, and more individuals experiencing it are seeking treatment. Alopecia areata, androgenetic alopecia, trichotillomania, traction alopecia, congenital alopecia, hair cycle abnormalities and tinea capitis conditions are frequent causes of alopecia in youngsters and teens. Alopecia manifests in different types, and each necessitates a distinct treatment approach. The review describes alterations in hair composition, biology, and the proteins involved in Alopecia, along with recent studies on treating this condition

Keyword: Alopecia, Types, Causes, Protein.

INTRODUCTION:

Alopecia is a condition that leads to hair loss or shedding in areas where hair is normally found, without causing scarring. Alopecia refers to a wide range of hair loss disorders impacting millions across the globe[1]. It can differ slight from thinning to total hair loss on the body and scalp. Alopecia can impact facial hair and can manifest in various areas of the scalp, beard, eyebrows, eyelashes, ears, and more. It involves hair loss in spots that vary from approximately 1 cm to extensive regions on the scalp[2]. Pregnant women might also encounter small areas of hair loss. Individuals with Alopecia might undergo spontaneous remission, but may also face recurring episodes. The regenerated hair's type, color, and texture may not necessarily match the original hair[3].

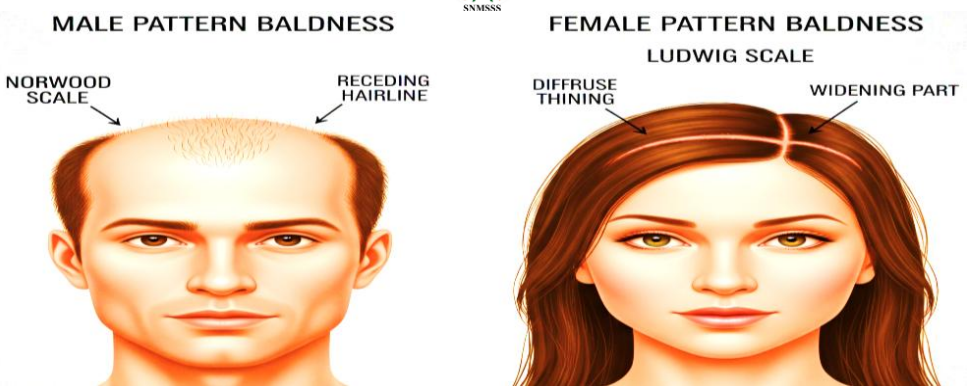

Figure 18.1. Male and Female Pattern of Baldness

Certain types of alopecia may be transient, while others are enduring and necessitate medical treatment. Genetic, autoimmune, hormonal, or environmental factors can cause the condition. Alopecia can be concentrated or widespread, short-term or long-lasting. It can impact individuals of all genders and age ranges[4]. Patients may experience considerable distress, reducing their quality of life. A comprehensive history, thorough physical examination, and targeted assessment are essential to identify the root cause, which will inform choices about the most suitable management for optimal results. Handling alopecia can be challenging. There are occasions when Alopecia responds to the treatment, although its efficacy is questionable[5].

TYPES[6]:

Figure 18.2. Types of Alopecia

SYMPTOMS[7]:

Figure 18.2. Symptoms of Alopecia

CAUSES[8]:

- Genetics – Inherited hair thinning, including male and female pattern baldness.
- Autoimmune diseases – Conditions such as alopecia areata, in which the immune system assaults hair follicles.
- Hormonal fluctuations – Discrepancies in hormones such as dihydrotestosterone (DHT).
- Emotional or physical stress can cause temporary hair shedding. Nutritional deficiencies – Insufficient vital vitamins and minerals such as zinc, iron, and biotin.
- Medical therapies – Chemotherapy and radiation can result in hair loss.
- Infections and scalp issues – Fungal infections, psoriasis, and dermatitis may lead to hair loss

TREATMENT[9,10]:

The approach to treating alopecia varies based on the kind and intensity of hair loss. Altering methods often consist of:

- Topical solutions – Drugs such as minoxidil promote hair growth.

- Oral medications – Substances such as finasteride inhibit the production of DHT (JAK inhibitors).
- Steroid injections – Utilized for alopecia areata to inhibit immune reaction (Corticosteroid).
- Hair transplant procedure – Revives hair by moving healthy follicles.
- Lifestyle adjustments – Reducing stress, enhancing nutrition, and utilizing mild hair care items.

RECENT RESEARCH AND STUDIES[11]:

- Stem Cell Therapy – Research indicates that treatments utilizing stem cells may help regenerate hair follicles and promote hair growth. This treatment utilizes stem cells, capable of regenerating new tissue, to activate hair follicles and enhance hair growth. Although it is not a remedy, it may assist in regrowing hair in certain situations.
- Antibody Treatments – Researchers are investigating antibody-focused therapies to accurately target molecular mechanisms associated with hair loss. These antibodies focus on particular immune system proteins that play a role in hair loss, providing a novel method for addressing this issue.
- Microneedle Patch Therapy– Scientists from Harvard and MIT have created a microneedle patch that administers immune-regulating substances to targeted regions, promoting hair regrowth without inhibiting the entire immune response.

CONCLUSION:

Alopecia is a prevalent immune condition affecting individuals of all ages at varying prevalence levels. The disorder impacts children more significantly than adults and seniors. This review showed that elements like mental health, anxiety, stress, and depression significantly contribute to alopecia and can also exacerbate the condition. Worldwide, the incidence of Alopecia stands at 2%, with hereditary characteristics or psychological factors identified as underlying conditions upon examination. However, upcoming research still needs to identify viable interventions and issues related to Alopecia

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Chapter - 20

Study on Malnutrition it's Cause and Treatment

Raghul Shree. S

ABSTRACT:

Malnutrition is a significant issue in developing and underdeveloped nations. A rising number of children and pregnant women are losing their lives due to insufficient nutrition in their daily diet. The importance of food's nutritional value during the growth phase is significant. The growth of the human body necessitates different forms of nutrition for proper development, acquired through a consistently healthy diet. Malnutrition is a frequent, often unacknowledged and insufficiently treated issue among hospital patients. Disease-related malnutrition results from decreased dietary consumption, malabsorption, heightened nutrient losses, or modified metabolic requirements.

Keywords**:** Malnutrition, Types, Impact, Treatment.

INTRODUCTION:

Malnutrition happens when a person receives insufficient or excessive nutrients, leading to health issues. It is specifically a lack, surplus, or disproportion of energy, protein, and other nutrients that negatively impacts the body's tissues and structure. Even after years of investment aimed at addressing this issue, India's rates of child malnutrition, which account for a significant portion of the global undernutrition burden, remain among the most concerning worldwide[1].

The Global Hunger Index (2022), determined by child mortality, child stunting, overall population undernourishment, wasting and India rank in 107th/121 countries. Released each year by Concern Worldwide and Welthungerhilfe, the Global Hunger Index investigates the existing data on hunger rates globally[2]. Here are the 10 nations with the highest rankings in 2025 -1. Somalia, 2. South Sudan, 3. Republique Democratique du Congo, 4. Madagascar, 5. Haiti, 6. Chad, seven. Niger, 8. Republique Centrafricaine, 9. Nigeria, 10. Papua New Guinea. At present, over 30 million children in the 15 most impacted nations are affected by this issue, with 8 million of them experiencing severe wasting

Malnutrition continues to be a significant global health issue, impacting countless individuals of all ages and economic statuses. It extends beyond hunger by including various

conditions that occur when the body lacks sufficient, balanced, or suitable nutrients. This can present in two primary ways: undernutrition, encompassing stunting, wasting, and shortages of vital vitamins and minerals, and overnutrition, associated with obesity and diet-related noncommunicable illnesses[3].

The effects of malnutrition reach well beyond personal health, resulting in hindered physical and mental development, lower productivity, and greater susceptibility to illness[4]. In spite of progress in healthcare and food production, malnutrition remains a challenge due to intricate issues like poverty, inequality, unhealthy eating habits, and restricted access to healthcare. Grasping the reasons, effects, and potential remedies for malnutrition is essential in tackling this complex challenge and fostering global sustainable development[5].

TYPES[5]:

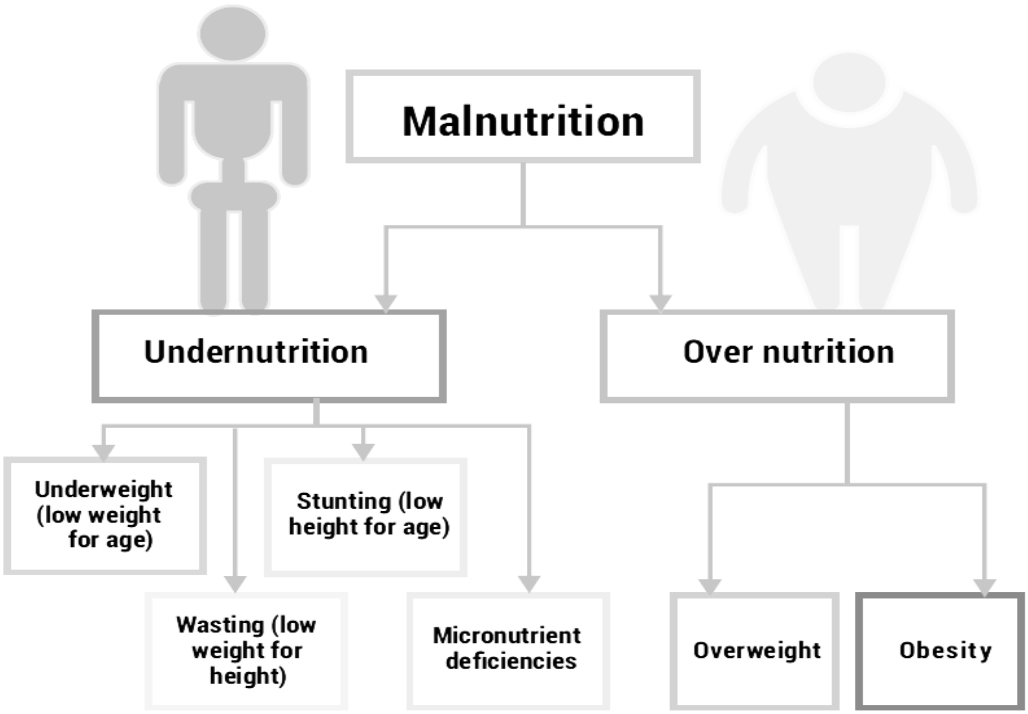

Figure 20.1. Types of Malnutrition

EFFECTS[6]:

Figure 20.2. Effects of Malnutrition

PREVENTION[9]:

Malnutrition can be avoided by eating a balanced, healthful diet. The following are the four main food groups for a balanced diet:

Table 20.1. Prevention of Malnutrition

Bread, rice, potatoes, and other starchy foods	Portion of the diet	Supply calories for energy
Milk and dairy foods	Vital sources of fats and simple sugars	Supply Lactose and Calcium
Fruit and vegetables	Vital sources of vitamins and minerals, fiber and roughage	For better digestive health
Meat, poultry, fish, eggs, beans and other non-dairy sources of protein	Building blocks of the body	Help in numerous body and enzyme functions

FACTORS AND CONSEQUENCES[7,8]:

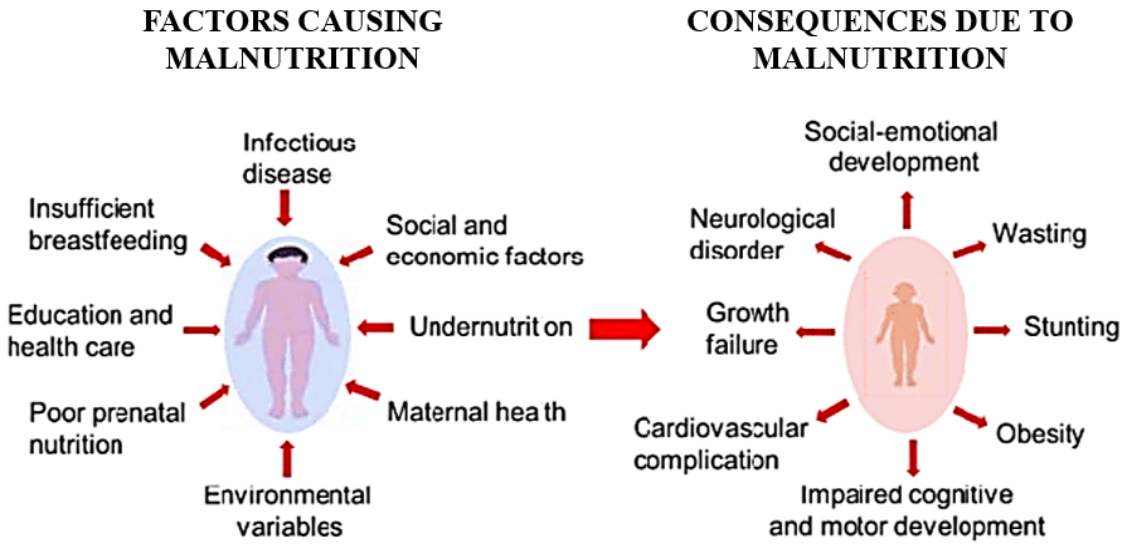

Figure 20.3. Factors and Consequences of Malnutrition

TREATMENT[10]:

Enhancing nutrition: Development assistance encompasses feeding practices for infants and young children, promoting breastfeeding, and interventions involving products such as ready-to-use therapeutic foods, micronutrient strategies, and vitamin supplements.

Detecting malnourishment: Measuring children is essential for recognizing malnutrition, which aids in understanding its effects on child health.

Micronutrient supplementation: The World Bank recommends addressing malnutrition with minerals such as iron, iodine, zinc, vitamins A and D, potassium, and magnesium to aid in restoring electrolyte balance.

Therapeutic foods: Foods high in lipids and soluble in water consist of: n-3 fatty acids, n-6 fatty acids, Niacin, Pantothenic acid, Folic acid, Vitamins (E, K, C, B1, B2, B6, B12) and Biotin utilized for the urgent feeding of those who are malnourished.

CONCLUSION:

Malnutrition, frequently neglected by healthcare professionals, is prevalent and impacts various physiological functions significantly. It is linked to higher rates of illness and death among hospitalized patients and greatly raises healthcare expenditures. Every physician must acknowledge that adequate nutritional management is essential for effective clinical practice. By tackling gaps in the education of all healthcare providers and significant advancements in nutritional care, leveraging clinical leadership can be achieved. By recognizing its origins,

symptoms, and lasting impacts, we can collaborate to develop enduring solutions that encourage healthy development and well-being for all, no matter their location

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Chapter - 21

DNA Fingerprinting in Today's Investigative Field

Rakshita. P

ABSTRACT:

DNA fingerprinting, one of the significant discoveries of the late 20th century, has revolutionised various investigative fields (Paternity, Maternity, Forensic testing, Disease identification, organ matching and so on). Numerous genetic alterations can be found using DNA fingerprinting. Every person on the planet may be identified at the molecular level thanks to an incredibly high degree of polymorphism in their DNA sequence, which they inherit from their biological parents and are the same in every cell of their body. Despite the fact that every human has 99.9% identical DNA sequences, there is enough variation in DNA to allow for individual identification. Before genomes and proteomics techniques were developed, this separation was based on physical traits.

Keywords: DNA, Samples, DNA Profiling, Investigative fields.

INTRODUCTION:

Sir Alec Jeffreys of the University of Leicester in the United Kingdom first revealed the DNA profiling technique in 1986 and now serves as the foundation for various national DNA databases[1]. The human body comprises 60 trillion cells, and each diploid cell in humans has 23 pairs of chromosomes (46 total), with half inherited from both parents. The complete DNA in a haploid cell consists of 3x10^9 BP. Approximately 99.9% of the DNA code is identical across all individuals. only 0.1% of it serves as the foundation for DNA profiling which is identified in the non-coding area. In total human DNA content only 3% acts as coding DNA remaining 97% act as non-coding DNA[2,3].

The significant quantity of non-coding DNA displays a uniqueness. It is made up of extensive sequences of consecutive repetitions of nitrogenous bases. Repetitive DNA makes up around 50% of the human genome. The count of repeats varies among individuals, and this characteristic is referred to as tandem repeat polymorphism, which is utilized in the DNA profiling procedure[4,5].

DNA profiling methods, founded on repetitive DNA sequences, have demonstrated immense significance, though the full application of this knowledge has yet to be fully realized[6]. A single hair, skin particles and a drop of blood can serve to detect DNA arrangements. It has a

numerous application in both legal and forensic contexts. Due to progress in forensics over the last forty years, DNA evidence has become one of the most dependable types of proof in a courtroom[7].

PROCESS INVOLVED IN DNA FINGERPRITING[8,9]:

The 11 essential steps in creating a DNA fingerprint are:

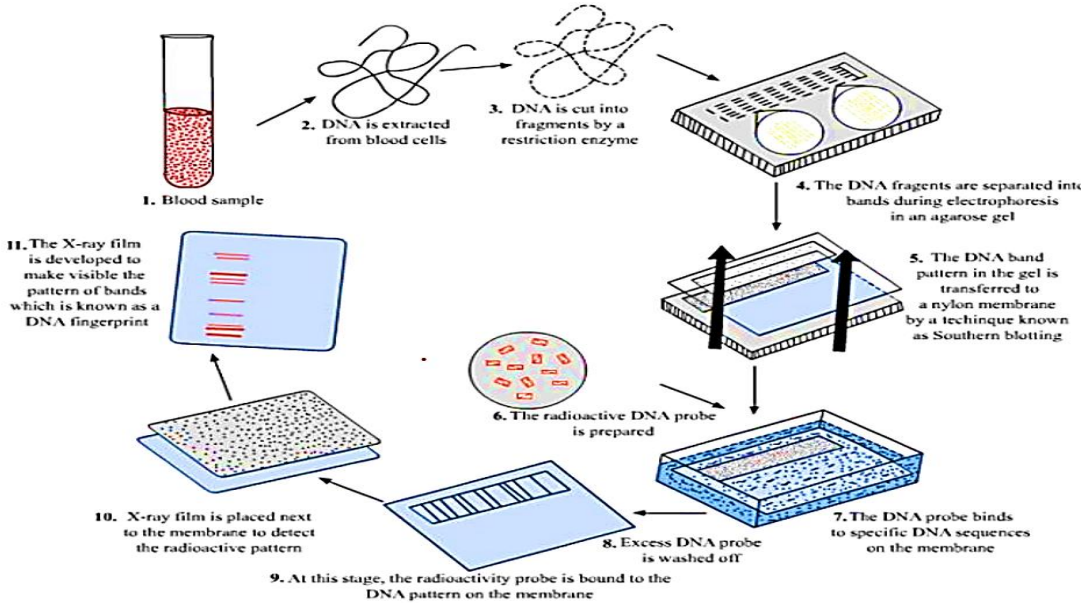

Figure 21.1. Steps involved in DNA Fingerprinting

APPLICATIONS OF DNA FINGERPRINTING INVOLVES[9-12]:

Figure 21.2. Applications of DNA Fingerprinting

1. Criminal Identification: Associates suspects with crime scenes by examining biological evidence such as blood, hair, or saliva, connecting them to serious offenses like murders and rapes.

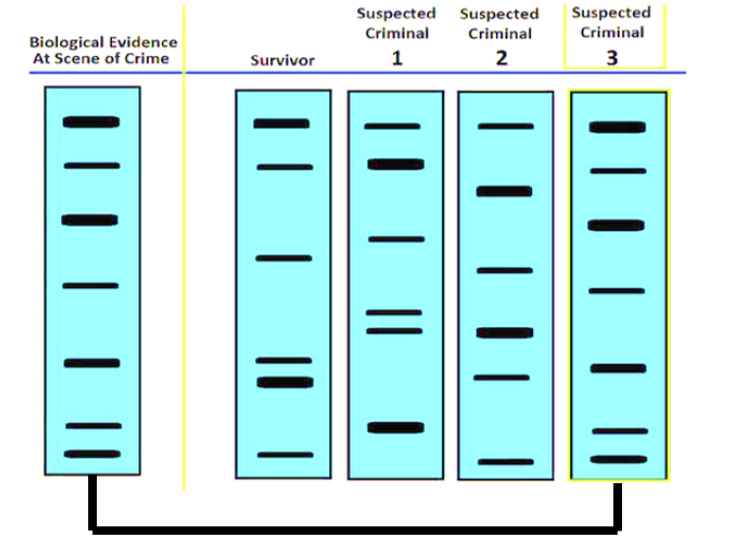

Figure 21.3. Criminal Identification in DNA Fingerprinting

In forensics, DNA profiling has become a fundamental aspect of criminal investigations. It aids in accurately identifying suspects while also being essential in demonstrating innocence, thereby acting as a protection against unjust convictions.

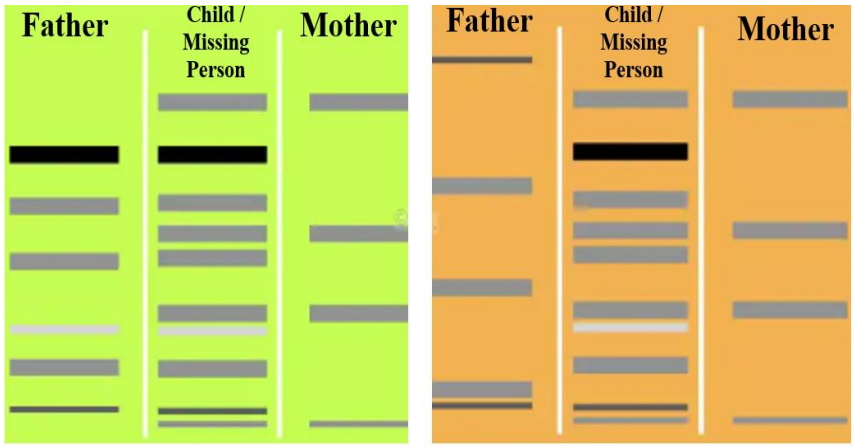

Figure 21.4. Identification of Child missing using DNA Fingerprinting

2. Paternity/Maternity Testing: Determines biological parentage by analyzing a child's DNA in relation to that of the mother and the possible father.

3. **Missing Individuals:** Recognizes unidentified bodies, frequently resulting from accidents, natural calamities, or criminal incidents.

In testing for paternity and maternity, this approach provides scientific certainty in situations involving contested parentage.

4. Identifying diseases: Important for diagnosing genetic conditions such as cystic fibrosis, thalassemia, huntington's disease and sickle cell anemia.

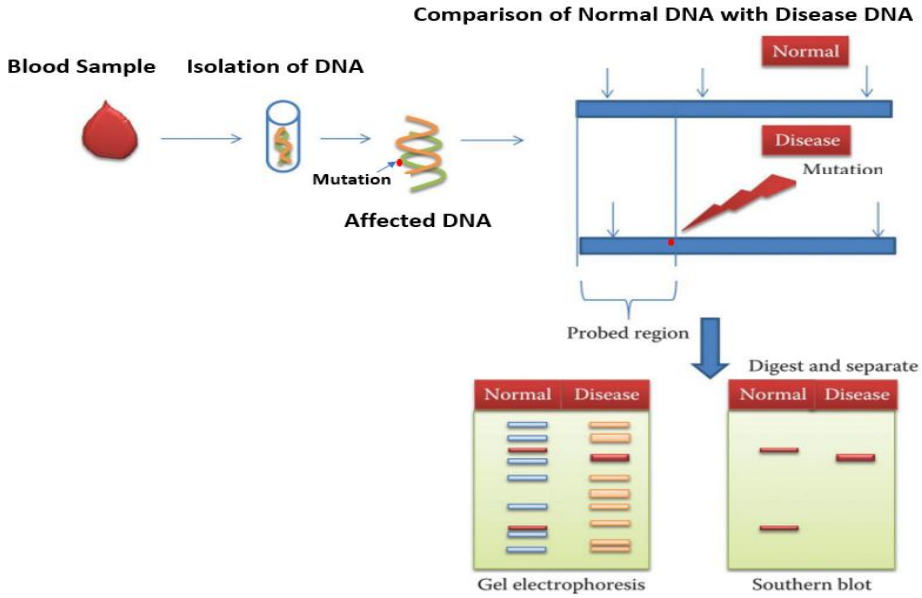

Figure 21.5. Identification Diseases using DNA Fingerprinting

In the field of medicine, DNA profiling has enabled early identification of genetic disorders, discovery of genes associated with diseases, and tailored medical treatments.

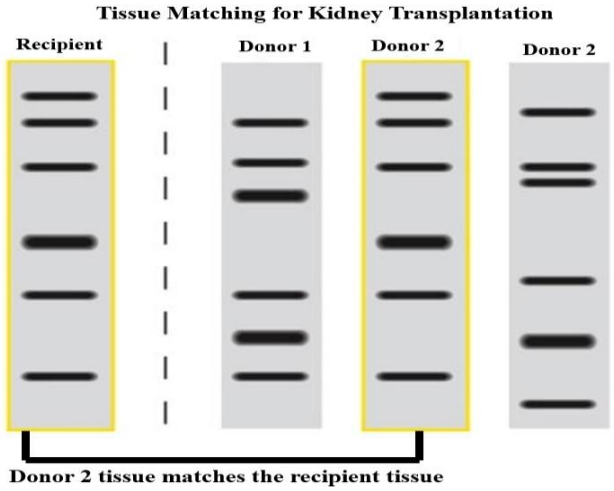

Figure 21.6. Identification Tissue matching using DNA Fingerprinting

5. Matching organs and tissues: It can be utilized to align the tissues of organ donors with

those of patients receiving transplants. Renal Transplant Surgery

In medical science, DNA profiling has enabled early detection of genetic diseases, the identification of genes associated with illnesses, and tailored medicine.

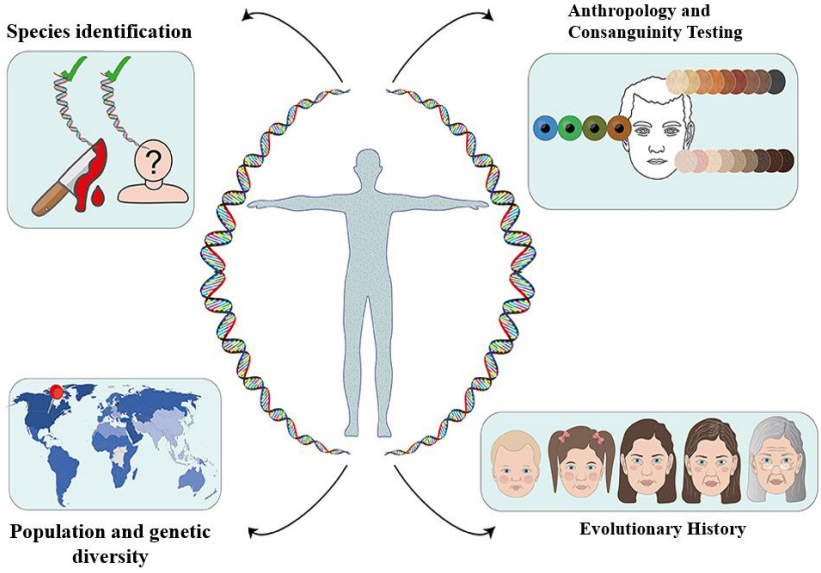

Figure 21.6. Identification Diversity using DNA Fingerprinting

6. Anthropology: Analyze ancient DNA samples to trace genetic connections and population migrations of humans across generations.

7. Evolutionary Background: Utilized in the examination of phylogeny and mapping the evolutionary background and connections among organisms.

8. Consanguinity testing: This tool can determine the level of relationship between individuals by indicating whether they have a shared ancestor.

9. Demographic and genetic variety: In forestry and wildlife management, DNA profiling aids in evaluating genetic diversity and structure among populations, essential for conservation efforts.

10. Species identification: This aids in recognizing animal and plant species, especially when dealing with small or degraded biological specimens.

CONCLUSION:

DNA profiling is among the most impressive accomplishments of contemporary science, as it has transformed how we identify, examine, and comprehend individuals genetically. This method offers a precise and trustworthy means of differentiating individuals by analyzing very specific DNA patterns. Its strength is rooted in the distinctiveness of each person's DNA, rendering it an essential technique in medicine, forensics, and research. In summary, DNA fingerprinting is more than a laboratory technique; it is a revolutionary scientific instrument that has changed law, medicine, and research. Its uses keep expanding with technological progress, guaranteeing that this method will stay a vital force in the future of science, justice, and human well-being

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Chapter - 22

A Review of New Approaches in Medicine - Gene Therapy

Tharun Kumar. V

ABSTRACT:

Gene therapy is recognized as the capacity for genetic enhancement through the rectification of altered (mutated) genes or precise modifications that aim at treatment. This treatment became feasible due to advancements in genetics and bioengineering that allowed for the manipulation of vectors to deliver extrachromosomal materials to specific cells. A central emphasis of this approach is the enhancement of delivery vehicles (vectors), primarily consisting of plasmids, nanostructures, or viruses. Viruses are frequently studied due to their ability to infiltrate cells and introduce their genetic material. Current vector systems can transfer genes into living cells (within the human body), but an optimal vector for gene delivery has yet to be discovered. Consequently, the existing viral vectors must be applied with substantial care in human situations. Additionally, the creation of new vectors is essential.

Keywords: Gene therapy, Mutated genes, Vector.

INTRODUCTION:

Gene therapy is a sophisticated medical approach designed to address or avert illnesses by directly altering the genetic material in a patient's cells. In contrast to conventional therapies that aim to alleviate symptoms, gene therapy addresses the fundamental origin of a condition at the molecular scale[1]. This method functions by substituting a defective or absent gene with a functional version, disabling detrimental genes, or adding new genes to assist the body in combating an illness[2].

The idea of gene therapy originated in the 1970s with the invention of recombinant DNA technology, but it gained substantial traction in the 1990s with the start of clinical trials. Currently, it stands as one of the most promising areas in contemporary medicine, particularly for genetic conditions like cystic fibrosis, hemophilia, muscular dystrophy, and specific cancer types. It is also being studied for acquired illnesses such as HIV/AIDS and heart diseases[3,4]. Gene therapy can be administered through two primary approaches:

- Somatic gene therapy - introducing functional DNA into the non-reproductive cells of a patient.
- Germline gene therapy - modifies generative cells and can be inherited by offspring[6].

Although germline therapy is contentious due to ethical issues, somatic therapy is extensively researched and has demonstrated promising outcomes[5].

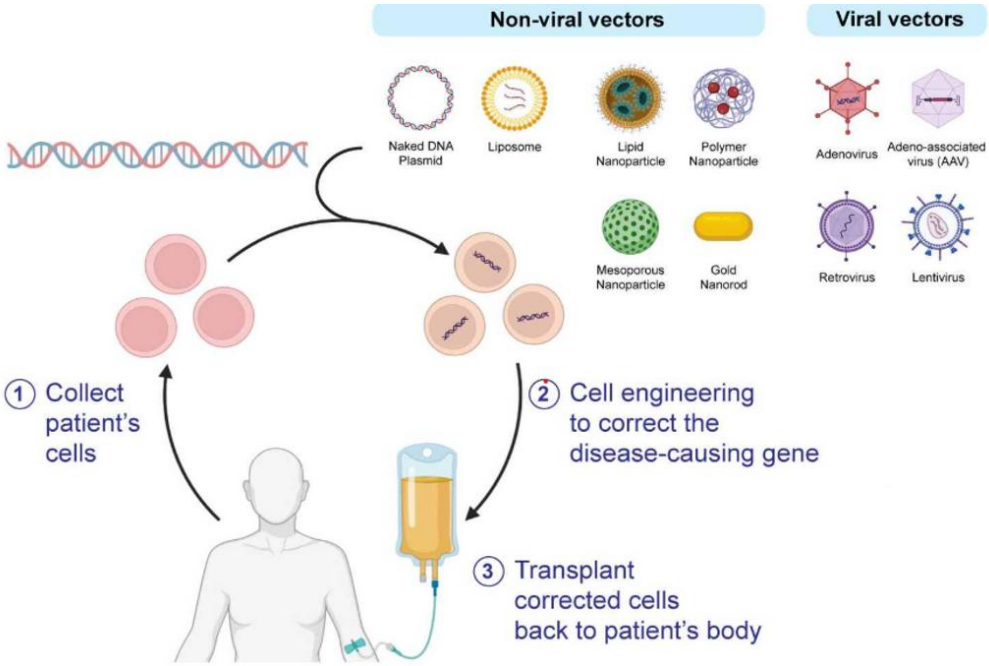
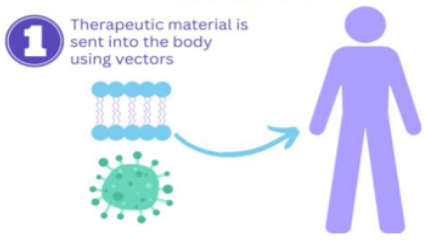
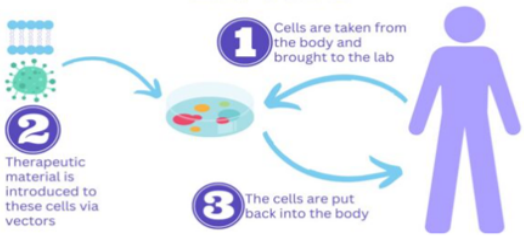


Figure 22.1. Steps involved in Gene Therapy

DELIVERY OF GENE THERAPY[7,8]:

There are 2 different ways of delivering the normal cell to patients to treat the disease.

IN-VIVO DELIVERY	EX-VIVO DELIVERY
To deliver genetic material specifically to an organ either through intravenous means or locally (for example, directly into the eye).	Procedure of extracting particular cells from an individual, modifying them genetically in a laboratory, and subsequently reintroducing them into the individual.
Functions with the assistance of a vector that directly delivers functional versions of a gene into target cells to address a missing or mutated gene.	Functions by genetically altering a patient's stem cells, which subsequently substitute for target cells that possess a defective or absent gene.
Targeted in vivo gene therapy will continue to evolve as scientists continue to refine methods of gene	Ex vivo gene therapy is most frequently applied to hematopoietic stem cells (HSCs)
	

GERMLINE THERAPY [9]:

Germline therapy entails altering the genes within germ or gamete cells, such as sperm or eggs. Germline therapy would thus be given during reproduction, as the altered gamete cells combine to create a zygote. In this manner, germline therapy modifies the genomes of generations yet to be born. Germline therapy is highly costly, which further restricts its practical application

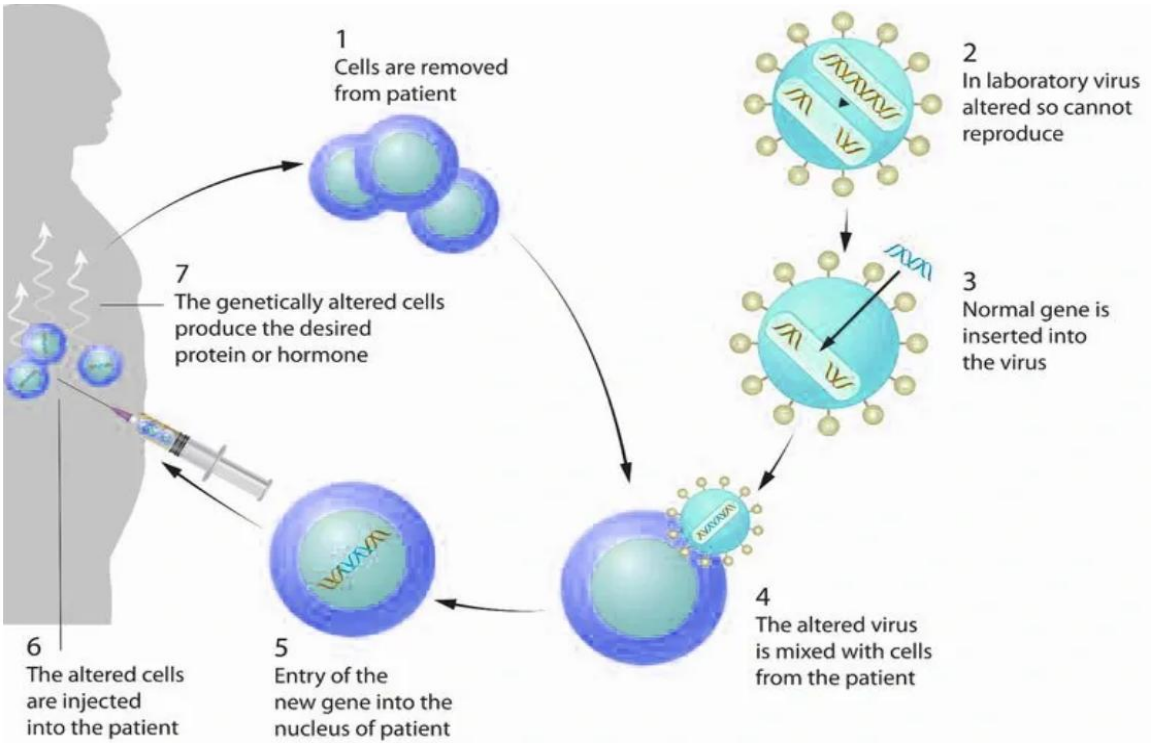


Figure 22.2. Steps involved in Germline Therapy

OBSTACLES IN GENE THERAPY[10]:

• Transporting the gene to the correct location and activating it.

• Evading the immune response,

• Ensuring that the new gene does not interfere with the functions of other genes,

USAGE OF GENE THERAPY[10]:

• It is utilized for substituting genes that lead to health issues.

• The approach typically eliminates the genes responsible for the problem.

• It assists the body in combating illnesses by introducing genes into the human system.

• This technique is used to address conditions like cancer, ADA deficiency, cystic fibrosis.

ASSESSMENT TO IDENTIFY MUTATIONS[11]:

Genetic testing can reveal which gene or chromosome contains a specific mutation. Genetic testing can aid parents in assessing their likelihood of bearing a children with a genetic disease if there is a familial history of such conditions.

AVOIDANCE OF MUTATIONS[11]:

- Applying sunscreen while outside and limiting your UV ray exposure.
- Wearing protective gear such as a mask and gloves when working with chemicals.
- Avoiding tobacco use.
- Consuming a nutritious diet and engaging in regular physical activity.

CONCLUSION:

Gene therapy signifies a groundbreaking advancement in medical science, providing the possibility of curing diseases previously deemed incurable. By addressing the underlying genetic causes of disorders, it offers a lasting and efficient solution instead of merely providing temporary symptom relief. The advancements made via continuous research and clinical trials have demonstrated significant potential in addressing genetic disorders, cancers, and specific viral infections. In the future, gene therapy might emerge as a standard treatment option, changing our approach to healthcare. It possesses the ability to both treat and prevent various genetic and acquired diseases. Ultimately, gene therapy represents a beacon of hope and creativity.

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Chapter - 23

Application of Bioinformatics in Research Field

Madhav Prasad

ABSTRACT:

Bioinformatics is a cross-disciplinary field of science that integrates computer science, mathematics and biology. Bioinformatics involves using biology evidence to handle genetic data, aiding in the analysis of plant genomes. The area of bioinformatics developed as an instrument to aid biological discoveries over a decade ago. The advancement of the Human Genome Project (HGP) remarkably and astonishingly boosted the amount of biological data. The capacity to collect, handle, process, analyze, and interpret data has become increasingly crucial. Scientists can use bioinformatics and computers to address it. This text presents the roles of bioinformatics, reviews the web tools and resources available in this field, and emphasizes its applications in agriculture along with its connections to other disciplines. Utilizing different bioinformatics tools in research which enables to analysis, retrieval, annotation, storage, and picturing of outcomes, enhancing the comprehension of biological systems in their entirety. This will aid in diagnosing and treating diseases for animal and plant health care

Keywords: Bioinformatics, Disease diagnosis, Database

INTRODUCTION:

Bioinformatics is an dynamic and emerging field that merges techniques, tools from biology and computer science to improve our understanding and interpretation of biological data. Bioinformatics applies computer science, programming, statistics chemistry, biology, physics, information engineering, data science and mathematics to interpret and analyse biological data[1].

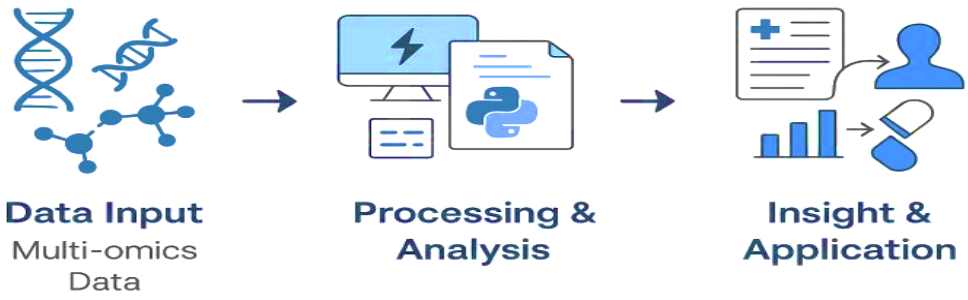

Figure 23.1. Stages of Bioinformatics

Bioinformatics provides insights into the data, enabling diagnosis for patients with rare diseases, monitoring and tracking infectious agents in populations, or determining the most effective treatment for cancer patients. It employs algorithms and software applications to examine biological data, including DNA sequences, protein formations, and gene expression information[2,3]. It also significantly contributed to the advancement of innovative technologies and instruments for examining genomic information. It has also been utilized to examine the functional importance of genetic variations and to locate possible targets for drug development[4]. The objective of bioinformatics is to reveal the vast biological information concealed within the extensive sequence, structure, literature, and other biological datasets.

DESIGNING DATABASES[5]:

This entails the arrangement, storage, and administration of biological data sets. Researchers can access the databases to review existing information and contribute new entries, such as the protein sequence data bank for molecular structures.

ANALYSIS OF DATA AND INTERPRETATION[6]:

The correct application of elements to evaluate the data and understand the outcomes in a biologically significant way. This encompasses DNA, RNA, and protein sequences, protein conformation, gene expression patterns, and biochemical pathways.

APPLICATION OF BIOINFORMATICS[7-10]:

Certain applications of Bioinformatics include:

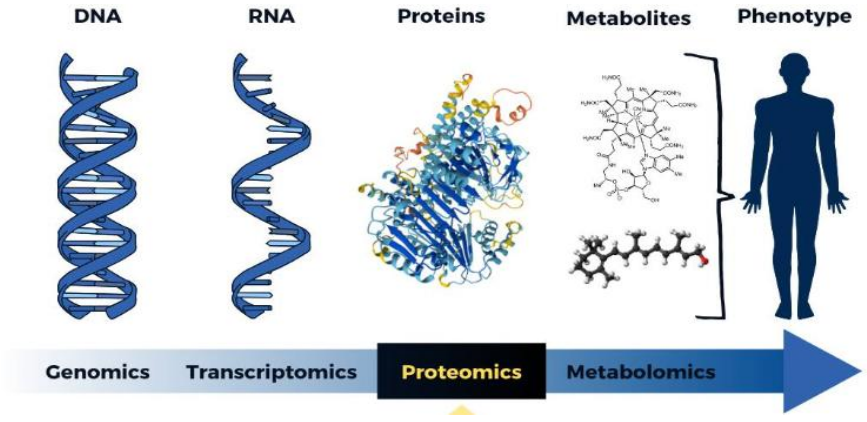

Figure 23.1. Applications of Bioinformatics

1. GENOMIC ANALYSIS:

Bioinformatics tools are used to handle the relationships amid DNA genes and sequences, the genetic variations and regulatory genome that may in traits or diseases. Genome-wide association studies employ bioinformatics methods involves in the identifying genomic diversity that could contribute to the development of composite traits.

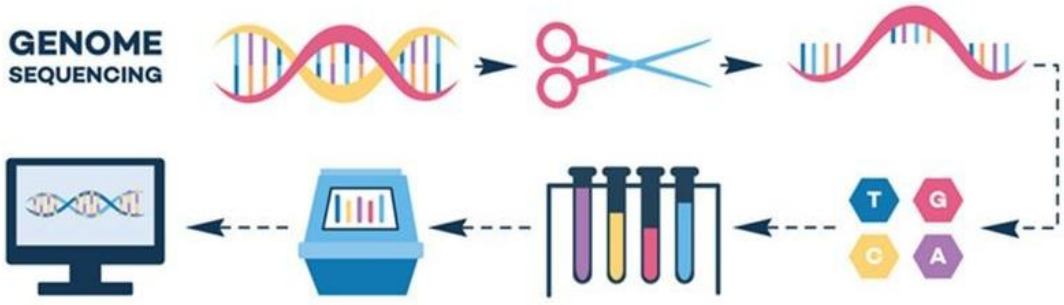

Figure 23.3. Genome Analysis

2. TRANSCRITOMICS:

Transcriptomics study utilizes RNA sequencing to examine gene expression patterns. Bioinformatics instruments enable the analysis of RNA-sequence data for various gene types, including alternative splicing events, differentially expressed genes and non-coding RNAs, offering insights into cells and its processes.

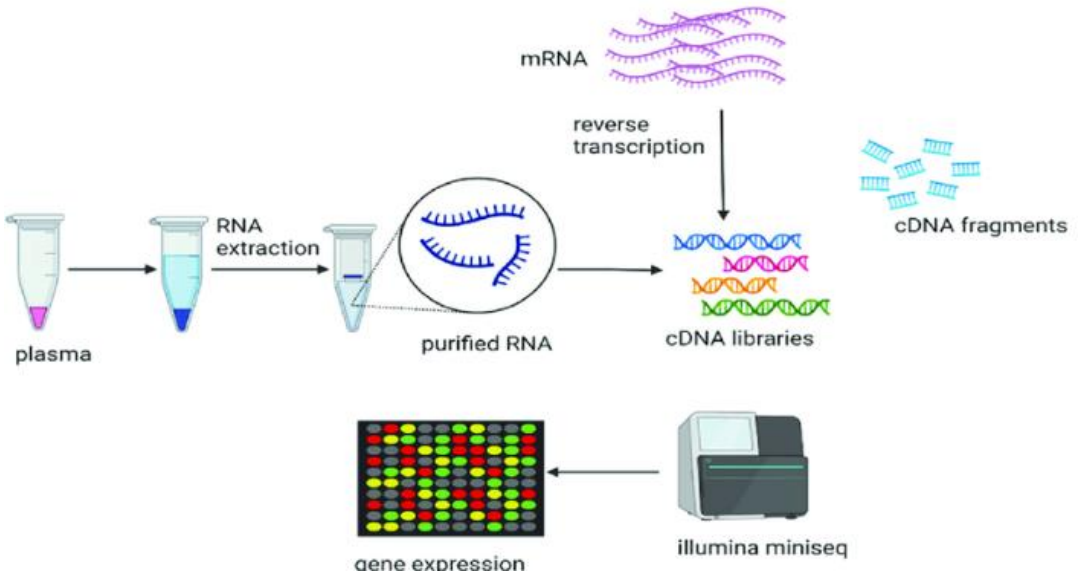

Figure 23.4. Transcriptomics Analysis

3. PROTEOMICS:

Domain of proteomics focuses on examining the function, structure and interactions of proteins within a genetic system. Bioinformatics tools are being utilised for various aims, including the quantification, identification and structural forecasting of proteins, the analysis of post-translational modifications and the exploration of protein-protein interactions.

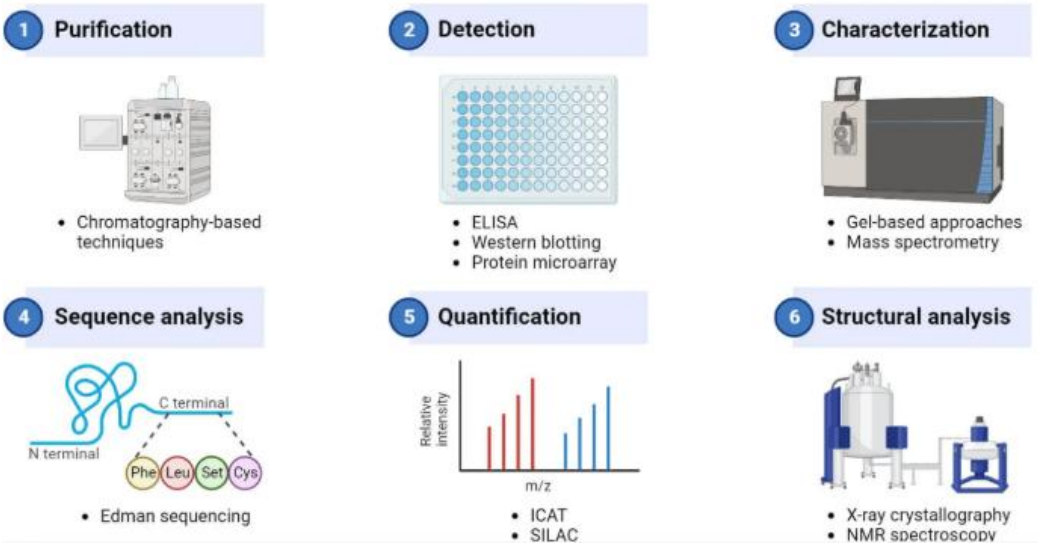

Figure 23.5. Proteomics Analysis

4. CLINICAL GENOMICS:

Research and clinics facilities utilise bioinformatics to analyse the genetics information of their patients, identifying the presence of genetic disorders, assessing disease risk, and recommending tailored treatments accordingly.

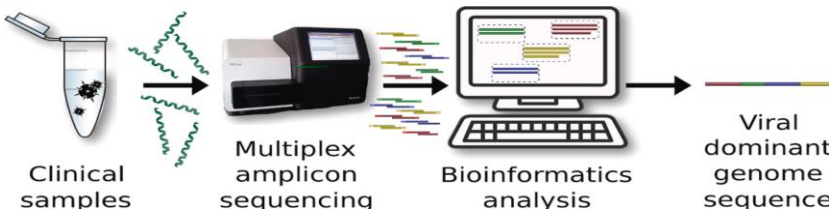

Figure 23.6. Clinical Genomics Analysis

5. STRUCTURAL BIOINFORMATICS:

The specific area of bioinformatics that focuses on predicting, analysing and modeling the structures of biomolecules, such as nucleic acids and proteins. Bioinformatics method

employs the predicting of protein structure, assessing docking and conducting computerized drug screening.

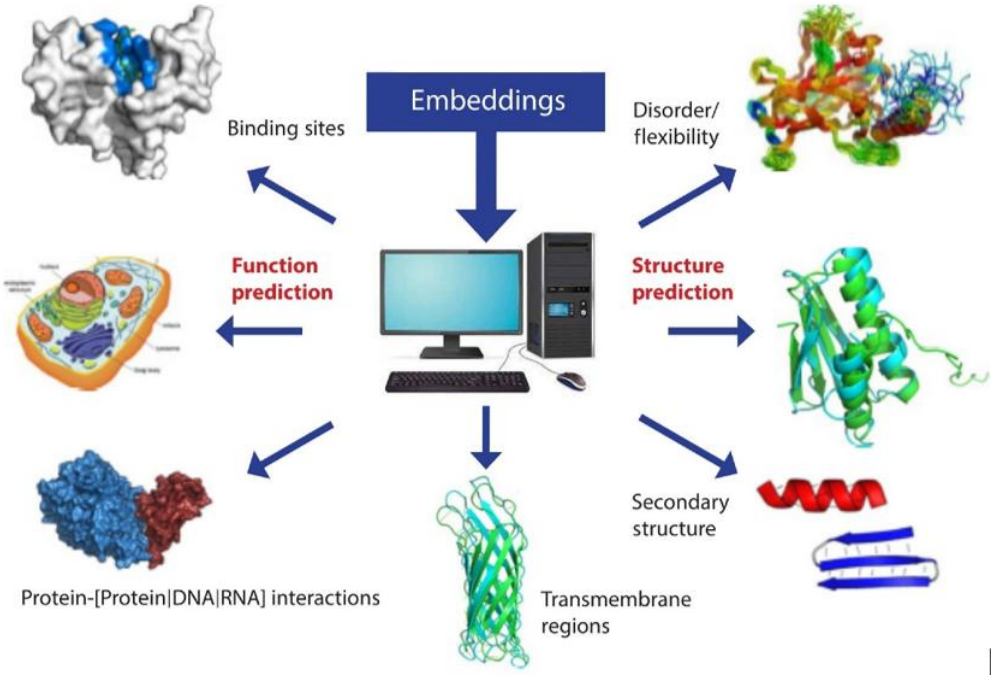

Figure 23.7. Structural Analysis

6. METAGENOMICS:

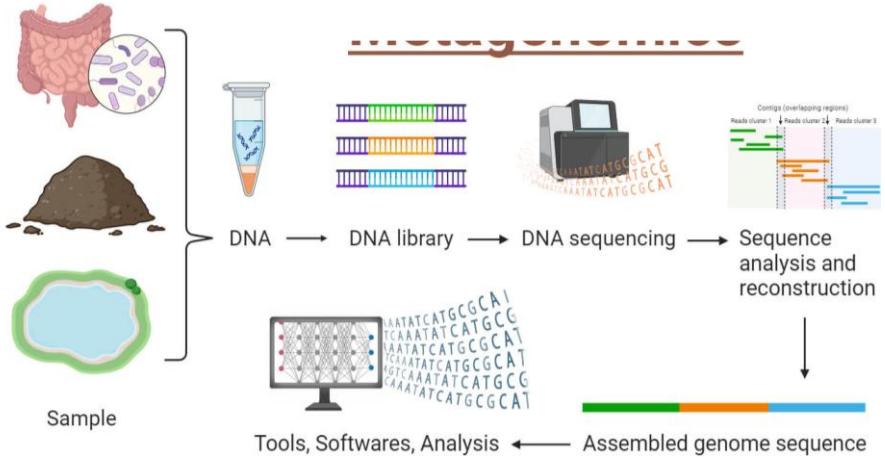

Figure 23.8. Metagenomics Analysis

Metagenomics involves examining the collective microbiological communities' genomes as they exist in environmental or soil settings. Utilizing bioinformatics methods, it is possible to analyze metagenomic data, examine different microbial species, forecast metabolic pathways, and explore microbial ecology.

CONCLUSION

Bioinformatics is an essential and evolving discipline that connects biology and technology, enabling the examination of biological data and promoting progress in research areas. Its capacity to analyze and comprehend vast datasets, combined with the advancement of advanced computational tools, has transformed our grasp of life and carries significant potential for the future of healthcare and biotechnology sectors. In summary, bioinformatics serves as a valuable resource for biotechnology firms to utilize for enhancing their comprehension of the data they examine. Bioinformatics has significantly influenced research and will undoubtedly keep doing so in the years ahead.

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Chapter - 24

A Comprehensive Analysis on Down Syndrome Diagnosis

Yashine. M. P

ABSTRACT:

Down syndrome is one of the most genetically intricate conditions that allow for human endurance beyond period, and it is the most common survivable autosomal aneuploidy. Down syndrome is one of the most prevalent conditions, incurring significant medical and societal expenses. Maternal age above 30 years elevates the chances of bearing a child with DS. DS is linked to various phenotypes such as Alzheimer's disease, congenital heart defects, Hirschsprung disease, leukemia and among others. While this complexity presents significant challenges in grasping the molecular foundations for each clinical feature of DS, it also offers chances to enhance comprehension of genetic mechanisms that drive the functionality and development of diverse tissues, organs, cell types and systems. Individuals with DS experience these phenotypes to differing degrees, making it crucial to understand the reasons behind this variation.

Keywords: Down syndrome, aneuploidy, Types.

INTRODUCTION:

Down syndrome (DS) is a genetic disorder that occurs once there is an extra copy of a chromosome 21. This additional chromosome can impact an person's physical traits, intelligence and overall growth. It also raises the chances of certain health issues[1]. This extra genetic information changes the development process and leads to various physical and cognitive traits.

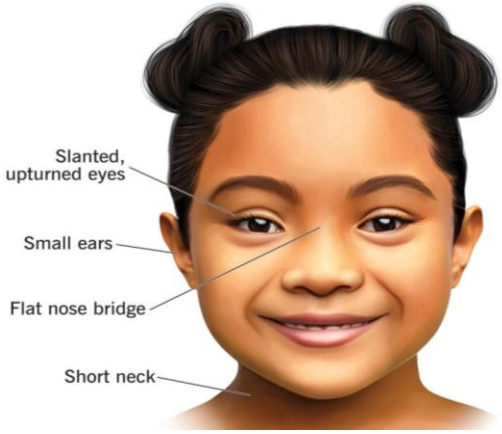

Figure 24.1. Down syndrome

The disorder is linked to different levels of cognitive deficits, distinctive facial characteristics, and a heightened likelihood of specific healthiness, such as respiratory complications ,congenital heart defects and auditory complications and thyroid ailments[2]. In spite of these obstacles, people with Down syndrome can have fulfilling and productive lives when they receive suitable medical care, educational assistance, and community involvement[3].

The occurrence of trisomy is posh by maternal age and differs among populations (reaching 1 in 319 - 1 in 1000 live births). DS exhibits considerable genetic variability and complexity in phenotype. In established nations, the typical life expectancy for the DS patient is 55 ages, and through various screenings and examinations, healthcare providers can identify Down syndrome either prenatally or postnatally[4]. Several factors influence it, but studies indicate that the likelihood increases if the mother is over 35 years old. Once individuals reach the age of 40, this number increases to approximately 12 out of every 1,000. This report examines the genetic foundations of Down syndrome, diagnostic techniques, prevalent health concerns, and current research[5]

CAUSES AND GENETICS[6]:

Normal human cell comprises 46 chromosomes, ordered into 23 pairs. An additional chromosome leads to Down syndrome.

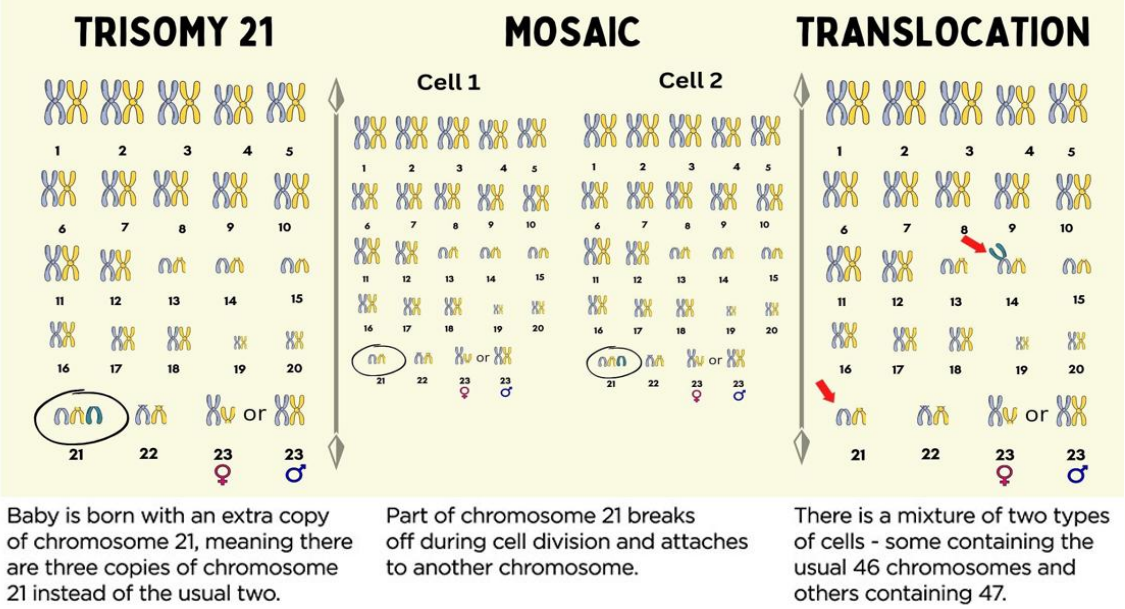

Figure 24.2. Different types of Down syndrome

Down syndrome arises when there are three copies of chromosome 21 rather than the typical two, leading to a total of 47 chromosomes. This additional chromosome 21 is found in every cell of the body.

DIAGNOSIS[7]:

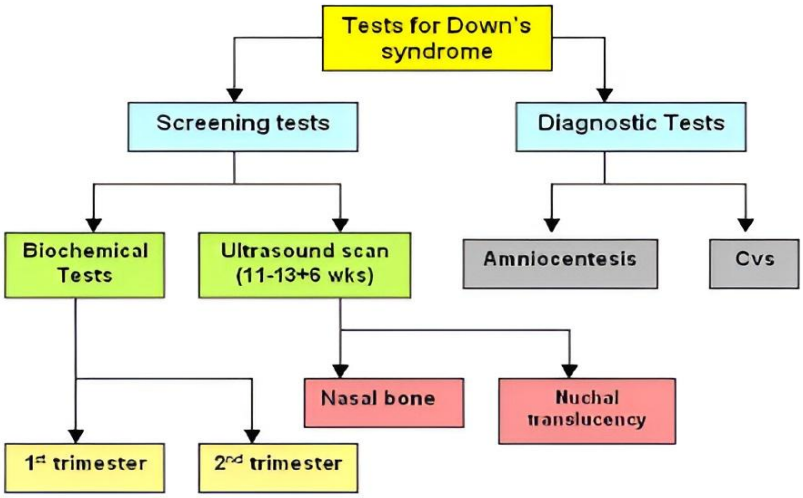

Figure 24.3. Types of Diagnosis in Down syndrome

COMMON HEALTH ISSUES[8]:

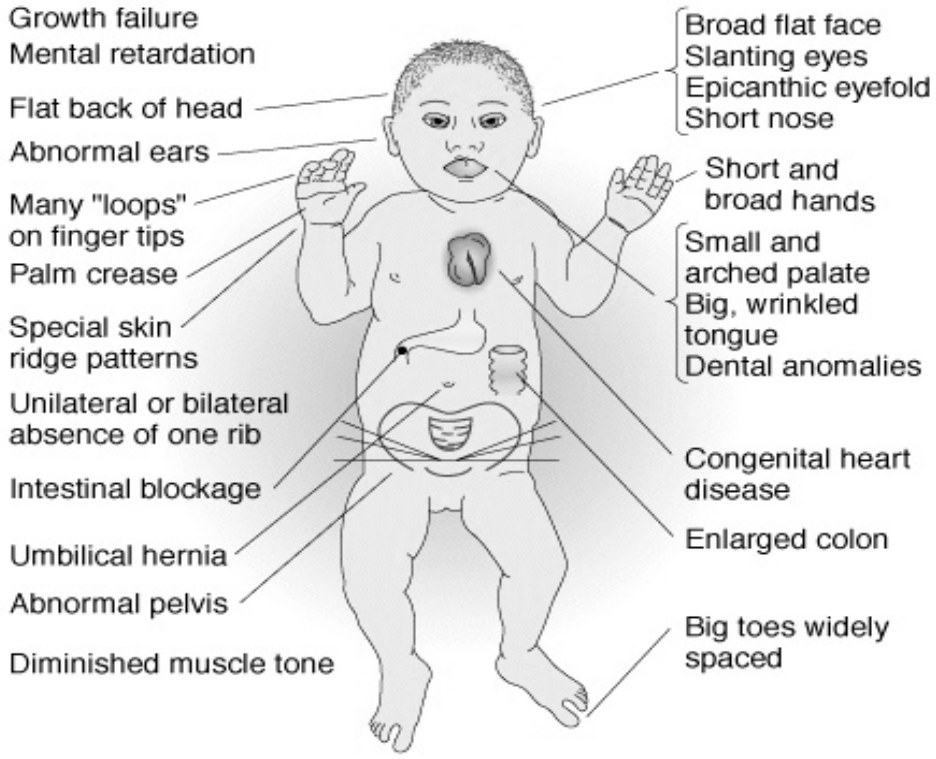

Figure 24.4. Down syndrome Symptoms and Signs

Individuals with Down syndrome might face various health problems, some of which are more prevalent or serious, while others might occur less often. An additional chromosome 21 can impact various systems in the body, increasing the susceptibility of individuals with Down syndrome to specific health issues.

TREATMENT[9]:

Figure 24.5. Treatment for Down syndrome

RECENT RESEARCH AND FUTURE OUTLOOK[10]:

Table 24.1 Recent and Future outlook of Down syndrome

Recent Research	Future Outlook
• Gene Modification and Chromosome 21 • Molecular Variants and Tailored Healthcare • Dysfunction of the Blood-Brain Barrier and Inflammation • Inhibitors of JAK for Autoimmune Disorders	• Progress in Early Detection and Genetic Treatment • Intelligent Automation in Diagnostic Processes • Obstacles in Regulation

CONCLUSION:

Down syndrome is a multifaceted genetic disorder impacting millions globally, affecting physical growth, mental capabilities, and overall well-being. Despite notable advancements, obstacles persist, particularly in guaranteeing fair access to healthcare, tackling ethical issues related to genetic interventions, and assisting individuals and families in practical environments. In summary, although Down syndrome is a lifelong condition, ongoing research, inclusive policies, and supportive communities can enable individuals with Down syndrome to enjoy healthier, more independent, and rewarding lives. The future promises significant opportunities as science, medicine, and society collaborate to overcome challenges and enhance results.

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Chapter - 25

A Review on Indian Medicinal Plants and Their Importances

Rathna Srinitha. K

ABSTRACT:

Medicinal plants serve as a crucial source of compounds with healing properties and continue to constitute an essential reservoir for discovering novel medications. The healing characteristics of plants are primarily attributed to the presence of active primary metabolites. Even though medicinal plants remain a crucial source for discovering new therapies, their scientific authentication is still insufficient due to selection, inadequate plant material, availability of bioactive compounds and application of appropriate high-throughput screening bioassays, in substantial amounts, and ultimately, their clinical trials and regulatory approval. For millennia, healing herbs have been utilized to address health issues, enhance flavor, preserve food, and avert disease outbreaks. In this review, we provided an overall summary of the therapeutic plants.

Keyword: Medicinal plants, Bioactive compounds, Pharmacological properties.

INTRODUCTION:

Traditional medicine practice is common in India, China, Sri Lanka, Japan, Thailand and Pakistan. Herbal remedies in Thailand, utilize legumes found in the Caesalpiniaceae, Fabaceae, and Mimosaceae families. China attributes 40% of traditional tribal medicines in their total medicinal consumption. It is estimated that in the mid-90s, the sales of herbal medicines generated over US$2.5 billion. In Japan, herbal medicinal products are more sought after than traditional pharmaceutical items. Medicinal plants are those plentiful in secondary biomolecules and possible source of remedies. Secondary biomolecules include flavonoids, alkaloids, coumarins, glycosides and steroids. Medicinal florae are used in recipes to treat common health issues such as diarrhoea, fevers, constipation, menstrual problems, hypertension, weak penile erection, low sperm count, dysentery and coated tongue. As a result, medicinal plants are viewed as home-based remedies in various regions of the nation. Medicinal plants serve as significant sources of nutrients and thus, they are suggested for their healing properties. In addition to their

medicinal applications, herbs are utilized in natural dyeing, pest management, food preparation, fragrances, tea, and more.

Table 25.1. Selected Medicinal Plants and its Properties

OCIMUM (Tulsi**)**

Scientific name	*Ocimum sanctum*
Uses	• Natural immunity booster due to Vitamin C and Zinc. • Used to cure mild fever, headache, or other simple ailments
Bioactive Molecules (Secondary metabolites)	• Eugenol methyl - anti-cancerous properties. • Eugenol - Stress-decreasing agent
Pharmacological properties	Enhance heart health, Anti-oxidant properties lower blood pressure, and Reduce hypertension.

ALOE

Scientific name	*Aloe vera and Aloe barbadensis*
Uses	• Used in flavouring liquors • Wound healer, Emollient or Moisturizer, in pharmacological and cosmetics formulations.
Bioactive Molecules (Secondary metabolites)	• Aloin and Aloe-emodin - analgesics, and possess antiviral/antibacterial properties.
Pharmacological properties	Anti-inflammatory, Antioxidant, Antimicrobial and Wound-Healing Effects

POPPY

Scientific name	*Papaver somniferum*
Uses	• Used as Psychoactive agents. • To reduce mental and nervous diseases
Bioactive Molecules (Secondary metabolites)	• Papaverine - smooth muscle relaxant. • Codeine - antitussive
Pharmacological properties	Antioxidant properties

ASHWAGANDHA

Scientific name	*Withania sominifera*
Uses	• Fruit, roots paste is used for curing ulcers, skin and bronchitis diseases. • Leave Paste used for treating inflammation in tubercular glands. • Used in Siddha medicines - rheumatism, cough debility from old age, dropsy. • Used in Unani medicines - general weakness. aphrodisiac, remunerative tonic, Diuretic, Hypnotic, Sedative, and restorative,
Bioactive Molecules (Secondary metabolites)	• Withaferin A - Antitumor properties, carbuncles • Withanin and Sominiferin - Anti-viral properties.
Pharmacological properties	Neuroprotective, Anti-Stress, Anti-Inflammatory, Immune-Modulating Agent

PERIWINKLE

Scientific name	*Catharanthus roseus*
Uses	For both ornamental landscaping and medicinal purposes - to treat diabetes, sore throat, skin infections,
Bioactive Molecules (Secondary metabolites)	• Raubasin - Hypertensive properties. • Serpentine - Antispasmodic properties. • Vincristine and Vinblastine - Cancer therapy • Vinblastine sulfate - Hodgkin's disease.
Pharmacological properties	Anti-ulcer, Neuroprotective, Antioxidant and Antimicrobial, Wound Healing

ISABGOL (PLANTAGO)

Scientific name	*Plantago ovata*
Uses	• Possessions of absorbing water - used an anti-diarrhoea medicine. • Treat chronic amoebic dysentery.
Bioactive Molecules (Secondary metabolites)	Rabinose, Rhamnose, Galacturonic Acid, Linoleic acid, Oleic acid, Linolenic acid.
Pharmacological properties	Antioxidant and Anti-inflammatory properties.

SENNA

Scientific name	*Cassia angustifolia*
Uses	• Least harmful and most consistent laxative agent. • In European countries consumed as the herbal tea • Used to treat acute constipation • Used to cleanse the colon
Bioactive Molecules (Secondary metabolites)	Sennosides A and B, Rhein-anthrone
Pharmacological properties	Anti-inflammatory and Anti-obesity

Glory Lily (Gloriosa)

Scientific name	*Gloriosa superba*
Uses	• Stimulating labor pains. • Useful in cases of gonorrhoea, chronic ulcers and piles. • Paste of the leaves is applied in the neck and forehead of children to cure asthma
Bioactive Molecules (Secondary metabolites)	Colchicine, Colchicoside, Isoperlolyrine - Anti-cancer.
Pharmacological properties	Antiperiodic, Antihelminthic, Antivenom.

RAUVOLFIA

Scientific name	*Rauvolfia serpentine*
Uses	• Roots become an essential part of Unani and Ayurvedic treatments in India • To treat ailments of the insanity, behaviour, maniacal, schizophrenia, etc.
Bioactive Molecules (Secondary metabolites)	Reserpine, Rescinnamine, Ajmalicine - Sympatholytic agent, Reducing blood pressure
Pharmacological properties	Antipsychotic, Sedative, Anti-Inflammatory Agent

SOLANUM

Scientific name	*Solanum Khasianum*
Uses	• Solasodine found in the plants, used as a substitute for diosgenine in the production of steroids. • Synthesis as the oral contraceptive tablets.
Bioactive Molecules (Secondary metabolites)	Solanine, Solasodine - Anti-proliferative effects
Pharmacological properties	Antioxidant, Anticancer, Anti-Inflammatory, Antimicrobial, Insecticidal

STEVIA

Scientific name	*Stevia rebaudiana*
Uses	Healthy substitute to sugar and henceforth consumed as an diabetic tonics.
Bioactive Molecules (Secondary metabolites)	Apigenin, Luteolin, Isoquercitrin, Kaempferol - Anti-Tumour activity.
Pharmacological properties	Antihyperglycemic, Antihypertensive, Antioxidant, Anti-Inflammatory Effects.

AONLA

Scientific name	*Phyllanthus emblica*

Uses	• Fruits are beneficial in haemorrhages, dysentery, jaundice, anaemia, cough dyspepsia, and. • Significant in Chavanaprash, Triphala and an excessive health vivacity restorer
Bioactive Molecules (Secondary metabolites)	Aonla, Gallic acid, Allagic acid is rich in Vitamin C
Pharmacological properties	Antioxidant, Anti-Inflammatory, Antimicrobial, Hepatoprotective, Anti-Diabetic Effects.

CONCLUSION

Medicinal plants are crucial, offering both well-known drugs and traditional remedies for numerous health issues, and their significance is rising with the heightened global demand. Although medicinal plants have historical and continued importance, they are currently threatened by overexploitation and extinction, requiring immediate conservation actions and sustainable harvesting methods. Ongoing research is crucial to confirm traditional practices and create safe, effective, and quality-regulated herbal medicines for wider public health advantages. The future of medicinal plants requires collaborative research, innovative scientific methods, and thorough conservation plans to realize their complete potential while safeguarding this essential biodiversity

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