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MARFAN SYNDROME: INSIGHTS INTO PATHOPHYSIOLOGY, DIAGNOSIS, AND TREATMENT STRATEGIES

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Marfan Syndrome, FBN1 gene, TGF-β pathway, beta-blocker, exome sequencing, drug therapy, ARB medicine, inflammation, fibrillin-1 protein

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Abstract

Marfan Syndrome is a single-gene dominant connective tissue problem mostly from harmful changes in the FBN1 gene, affecting the structure of fibers outside cells and proper elastic fiber function. These gene changes affect the TGF-β pathway, causing unusual tissue structure changes and increasing over time heart, eye, and bone problems. Population studies show that Marfan syndrome happens in about one in five thousand to one in ten thousand people worldwide, with nearly one-third of cases caused by new gene changes, showing variable gene effects and lots of differences in features. Visible traits go from mild bone problems to serious heart and blood vessel issues like bigger aorta base, artery bulge, and artery tear, which are still the main signs of sickness and risk of dying. Latest progress in detailed gene sequencing has increased the range of FBN1 gene changes, like hidden gene areas and mutations that change gene processing, helping update mutation categories and the link between genes and features. These findings show a move away from only watching blood vessels to complete heart muscle monitoring in Marfan syndrome care. Treatment plans have developed past beta-blocker drugs and ARB medicines, along with current studies on TGF-β blocking drugs, DNA-changing methods, and medicines affecting tissue structure. Some types of FBN1 gene changes affect the chance and speed of enlargement of the aorta base. Marfan disorder also has heart valve problems, like tricuspid and mitral valve bulge, and the mitral valve problem can be an early sign of serious childhood heart disease. Not related to heart valve problems, recent studies also show internal heart muscle problems and a higher chance of irregular heartbeats in the lower heart chambers. Heart ultrasound is done to find the disease, and new methods like the aorta base measurement help check children early. To see mitral valve separation and heart muscle scarring, heart MRI is very important. Because of the close connection between genes and features, DNA testing helps in finding high-risk cases and confirms the disease. Preventive aorta surgery using set limits, beta-blocker or ARB medicine, and monitoring for life are all included in treatment. This article shows that to improve results and well-being for people with Marfan disorder, complete heart treatment needs teamwork from different specialists and careful monitoring for all types of heart and blood vessel problems.

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INTRODUCTION

Marfan syndrome (MFS) is a body-wide connective-tissue condition that mostly happens due to harmful changes in the FBN1 gene, which makes an important protein outside the cells that is needed for forming microfibrils and keeping the body tissues flexible (Chen et al., 2022).

Fibrillin-1 fibers work like support structures for elastin layering, giving pulling strength to tissues. Damage to these fibers reduces tissue function and increases the risk of problems in multiple body systems (Milewicz et al., 2021). Marfan syndrome shows a dominant inheritance style, and one altered FBN1 gene copy is enough to cause the disease features. However, nearly one-third of cases occur due to new mutations, leading to random cases even in families with no previous history (Huang et al., 2025).These genetic changes cause clear differences in disease features, where bone, eye, and heart problems show to different degrees in people with similar gene changes (Zodanu et al., 2024; BMC Med Genomics Expert Panel, 2024). Studies on the general population show that Marfan syndrome occurs in about one in five thousand to one in ten thousand people worldwide, although missed diagnosis and partial showing of disease signs can hide the actual number of cases (Klemenzdottir et al., 2024).People who have increasing aorta size might need preventive surgery, because treatment with medicine only usually cannot stop blood vessel changes over time (Milewicz et al., 2021).Even though bone features like being tall, chest problems, curved spine, loose joints, and long fingers are some of the first visible signs, they do not have exact disease indication and can be similar to other connective tissue problems (Chen et al., 2022; Zodanu et al., 2024).

On the other hand, eye problems, especially lens dislocation, happen in about half of people with the condition and are still a clear sign of FBN1-related disease (BMC Med Genomics Expert Panel, 2024; Chen et al., 2022).How Marfan syndrome works has developed past just a problem with protein structure; recent studies show that uncontrolled TGF-β activity, tissue matrix changes, enzyme activity that breaks matrix, and smooth muscle cell loss are the main causes of blood vessel weakening (Zodanu et al., 2024).Progress in modern gene sequencing has found hidden, same-code, and splicing-changing gene changes in FBN1 that were missed by standard exon tests, broadening the types of cases that can be diagnosed (Sabahi Zadeh et al., 2025).New research have found that internal heart muscle problems, not connected to aorta enlargement, could be a separate cause of trouble with exercise and weak heart in children and adults with Marfan syndrome (Huang et al., 2025). Figure 01 shows checking multiple body parts rather than focusing only on the aorta, especially in young people who may develop worsening heart muscle disease (BMC Med Genomics Expert Panel, 2024).Using together genetic and medical monitoring lowers risk of dying and helps quick medical actions like surgery to replace the base of the aorta (Milewicz et al., 2021).So, this article tries to combine the gene structure, molecular processes, range of features, ways to diagnose, and new therapy approaches in Marfan syndrome, giving doctors and scientists a current plan based on research (Judge et al., 2021).Marfan syndrome is a genetic disorder that affects the body's connective tissues, which normally help hold organs and structures together. Because of this, many parts of the body are affected. People with Marfan syndrome often have a tall and thin body shape, with unusually long arms, legs, and fingers. Their joints are usually very flexible. The condition can also affect the heart and blood vessels, especially the aorta, making it weak or enlarged. Eye problems such as movement of the eye lens and poor eyesight are also common. These changes in body structure and organ function mean that people with Marfan syndrome need regular medical checkup (Huang et al., 2025)

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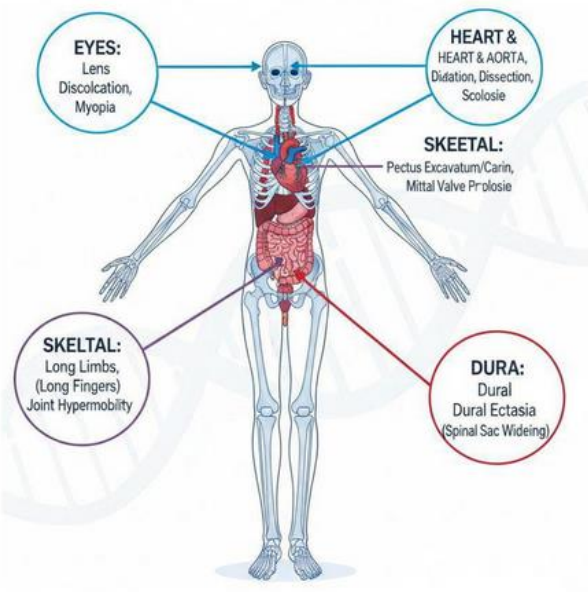

Fig. 01: Marfan syndrome connective tissue disorder

Prevalence of Marfan Syndrome

Marfan Syndrome (MFS) is an uncommon but important inherited disorder that affects the body s connective tissues. Current numbers show that it happens in about 1 in 5,000 to 1 in 10,000 people worldwide (Judge & Dietz, 2005; Loeys et al., 2010). Big studies on many people show that the way doctors identify MFS can change how common it looks. For example, a large study in Denmark found that about 6.5 people per 100,000 have MFS (around 1 in 15,500), and about 0.79 new cases are found per 100,000 people each year. This shows that new cases appear steadily in all age groups (Groth et al., 2015). Studies that only look at signs often report lower numbers because some people only have small symptoms, like long bones, eye troubles, or small heart problems, without meeting all the Ghent rules for diagnosis (Radonic et al., 2011).

The updated Ghent rules in 2010 focused more on a bigger aorta (Z-score ≥2) and harmful changes in the FBN1 gene. This helped doctors identify MFS more correctly and avoided mix-ups with other similar conditions, like the MASS phenotype, mitral valve prolapse, and Loeys Dietz syndrome (Loeys et al., 2010). After this change, the numbers became lower but more accurate, especially in heart clinics where wrong identification used to happen (Arnaud et al., 2017).

MFS does not prefer any gender or ethnic group. Both males and females are affected equally, and cases have been found in Europe, North and South America, South and East Asia, and Africa (Arbustini et al., 2014). Differences between places are mostly because of how easy it is to see a doctor, awareness about MFS, and availability of gene testing, not because of biology (Aftab et al., 2020). MFS seems more common in children because they often show bone and eye problems early, which brings them to doctors through bone or eye clinics (Lacro et al., 2014). But children who do not yet have a bigger heart may stay undiagnosed until teenage or young adult years, especially if they only have long limbs, very bendy joints, or a small curve in the spine (Radonic et al., 2011). In many countries, doctors check the aorta only if they think there might be a problem, which can delay identification (Lacro et al., 2014).

New gene tests have helped find more cases. Advanced DNA testing like next-generation sequencing (NGS) and whole-exome sequencing (WES) can find harmful FBN1 changes that older tests often missed, including hidden or unusual gene changes (Rommel et al., 2020). This helps correctly identify people who were unclear cases before.

In the past, most deaths from MFS without treatment were due to serious heart artery problems, like aortic

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aneurysm, dissection, or rupture. So missing cases is a big health danger (Hiratzka et al., 2010). Today, more awareness, early medicine, and preventive heart surgery have improved survival a lot. Family testing and regular future checkups also find relatives who were not diagnosed before, which increases the number of known cases over time (Lacro et al., 2014). Overall, the real number of people with Marfan syndrome is probably higher than reported. This is because less serious cases go hidden, gene changes may not show signs, many people cannot get gene tests, and family members are sometimes counted differently after a relative is diagnosed. Marfan syndrome can cause the fingers and toes to become very long and thin. The joints in the hands and feet bend easily, which may lead to weak control during movement. Many people develop flat feet because the tissues supporting the foot arch are not strong. Hand strength may be low, making it harder to grip or hold objects. Altogether, these effects can make everyday use of the hands and feet less comfortable (Lacro et al., 2014).

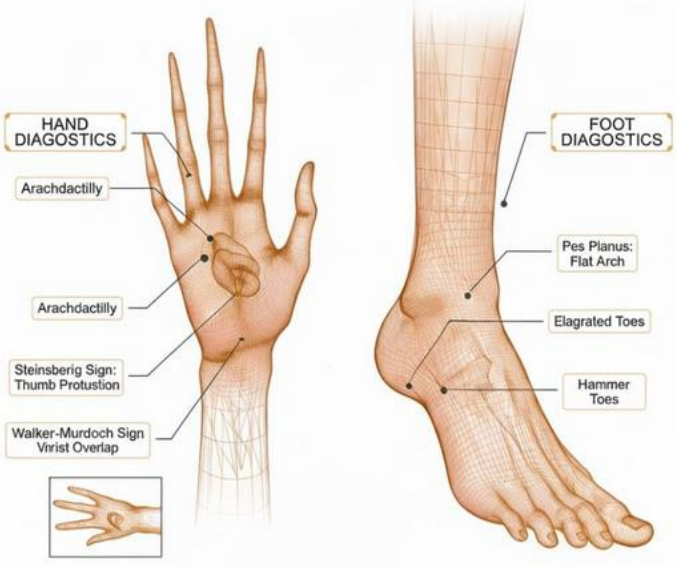

Fig. 02: Marfan Syndrome effect on hand and feet

Genetic basis and molecular mechanism of Marfan Syndrome

Marfan Syndrome (MFS) happens mainly because of damaging changes in the FBN1 gene on chromosome 15q21.1. This gene makes a protein called fibrillin-1, which is very important for making tiny fibers called microfibrils that give strength and support to the tissues in our body (Cheng et al., 2023). These microfibrils act like a framework to help another protein, elastin, form properly in the main artery (aorta), ligaments, eyes, and bones. The gene changes (mutations) in FBN1 can be of different kinds, such as missense, nonsense, frame shift, splice-site, or even hidden changes in DNA parts called introns or silent regions. Modern DNA tests like next-generation sequencing (NGS) and whole-exome sequencing (WES) can find these changes and make diagnosis easier (Takeda et al., 2023). Among all mutations, those affecting cysteine residues in special regions called calcium-binding EGF-like (cb-EGF) domains are the most harmful. They stop fibrillin-1 from forming properly, weaken the microfibrils, and damage the connective tissue structure (Cheng et al., 2023).

The problem in MFS is not only about weak fibers. Fibrillin-1 also controls a chemical signal called TGF-β by holding it in the space around cells (extracellular matrix). When fibrillin-1 is not working, TGF-β becomes too active. This causes wrong changes in tissues, death of blood vessel muscle cells (VSMCs), and more activity of enzymes (MMP-2 and MMP-9) that break tissue, which can damage the aorta and lead to bulges in the artery (aneurysms) (Zhou et al., 2021).Studies of aorta tissue from MFS patients show broken elastic fibers, build-up of jelly-like material, and

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loss of blood vessel muscle cells, showing that both structure and chemical signal problems happen together Hori et al., 2022). New studies looking at individual cells also found inflammatory blood vessel cells with overactive TGF-β and other chemical signals like interleukin-1β, suggesting that inflammation adds to tissue damage and blood vessel problems (Zhou et al., 2021).In simple words, MFS happens because of weak fibrillin-1, too active TGF-β, tissue damage, blood vessel cell death, and inflammation. Using modern DNA tests, tissue studies, and lab experiments, scientists now understand why MFS affects many parts of the body, why symptoms are different in each patient, and why finding the gene problem early is very important for care and treatment (Takeda et al., 2023).

Clinical Phenotype Spectrum of Marfan Syndrome (MFS):

Marfan Syndrome is a condition that affects the body's connective tissues and can show up in many different ways, especially in the heart, eyes, and bones(Chen et al.,2025) People with this condition can show many different symptoms, from small changes in their bones to dangerous aortic issues, and these differences mostly depend on the specific FBN1 mutation involved. (Bernhardt al., 2008,) .

4.1. Cardiovascular Phenotype:

Heart and blood vessel problems are the most serious and dangerous issues seen in Marfan Syndrome. A common and serious feature is the swelling of the aortic root in the sinuses of Valsalva, which can lead to life-threatening issues like aortic aneurysm, dissection, or rupture. .When the aorta continues to enlarge, preventive surgery to replace the aortic root is usually needed to stop serious problems from occurring. (T Chernof et al 2020). Problems like mitral valve prolapse, its leakage, and tricuspid valve prolapse are frequently seen and can play a role in developing heart failure later on. (Bibli, S. I.et al., 2020). Researchers have found that the heart muscle can be impaired on its own, and this can limit exercise capacity and contribute to early heart failure in people of all ages. (Razvi et al., (2023).

4.2. Ocular Phenotype:

The eyes are also commonly affected in Marfan Syndrome. The clearest sign is a shifted or dislocated lens, seen in 50 60% of patients, usually upward and to the side, caused by weak supporting fibers. (Jamieson, R. V.et al (2019).). Many patients also have myopia, a flattened cornea, or early cataracts. Glaucoma and retinal detachment occur less often, usually in those with more serious eye issues. (Basnett, S et al. (2013) The presence of ectopia Lentis in combination with other systemic features remains one of the most powerful indicators of MFS in clinical diagnosis.

4.3. Skeletal Phenotype:

Skeletal problems usually appear early but aren t always specific. Patients often have tall stature with long arms and legs, long fingers, chest deformities, curved spine, loose joints, and a high-arched palate. (Grill, F. et al(2013) . Patients may also show wrist and thumb signs, a lower upper-to-lower body ratio, and flattened arches in the feet. Marfan Syndrome can look very different from person to person, with some showing mostly skeletal problems and only minor heart issues, highlighting its variable nature. (Braverman et al 2021.

4.4.Pulmonary and Other Systemic Manifestations:

Lung and skin problems are less emphasized but still matter. Weak connective tissue and apical blebs can cause the lung to collapse unexpectedly. (Lewis, P. N. et al., 2025). Sleep apnea is more common in tall patients with chest abnormalities. Stretch marks on the skin and a stretched dural sac are also seen, and the dural changes can sometimes result in ongoing back pain. (Singh, N.et al (2025). In rare cases, nervous system issues like headaches and cerebral aneurysms have been seen.

4.5. Pediatric vs Adult Phenotypes

Marfan Syndrome signs evolve with age. In pediatric patients, long arms and legs, spinal curvature, and lens dislocation are often seen, while aortic problems may be silent, delaying detection of heart issues. (L. T., Lacro.et al 2025) Adults with Marfan Syndrome often experience growing aortic root size, valve dysfunction, and arrhythmias, while skeletal issues from childhood may continue or worsen over time. (Hoyos,et al.,2025)

4.6. Genotype Phenotype Correlations

The type of FBN1 gene mutation can affect the way the disease presents. Mutations causing lower fibrillin-1 levels tend to cause early and severe aortic issues. (van

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Der Hulst et al 2025) Dominant-negative missense mutations, particularly involving cysteine, tend to cause severe skeletal and eye features- (Devereux et al.,2025). This underlines the importance of genetic testing predicting risk and tailoring surveillance.

5.Diagnosis of Marfan Syndrome (MFS)

Finding out Marfan Syndrome (MFS) is difficult because its symptoms can change a lot, from small bone problems to very serious heart and blood vessel problems. (Limongelli, et al 2024). Therefore, it is not based on just one symptom, but on a mix of full body check-ups, scans, and gene tests. The updated Ghent rules (2010) are now seen as the best guide in the world. (Ali Akbar, et al., 2025) .They try to make diagnosis more accurate, avoid mistakes, and tell Marfan Syndrome apart from similar disorders like Loeys-Dietz syndrome, Ehlers-Danlos syndrome, and MASS (Clinical Genome Resource FBN1 Variant Curation Expert Panel, 2024).

5.1. Systemic Scoring , Multiorgan Evaluation

The systemic score is a well-structured formatted list assembling the skeletal, dermatologic, and cardiovascular characteristics. (Marelli, et al 2023) .Each sign gets points depending on how specific it is, and a score of 7 or more shows enough body system involvement for diagnosis when added with other rules: (Salles Rosa Neto et al., 2024). This point system lets doctors check body signs clearly and makes diagnosis more accurate, especially in unclear cases or in children. Overall, systemic scoring is a valuable tool for standardizing evaluation, ensuring thorough assessment, and aiding in early identification of MFS. Retrospective outpatient-cohort study.

5.2. Revised Ghent Criteria

The updated Ghent rules focus on heart and eye problems as the main parts of diagnosis, instead of just looking at bone problems like in older rules. (BMC Pediatrics Early-Onset Marfan Syndrome Collaboration, 2023). A bulging or tear in the main heart artery, lens dislocation in the eye, harmful FBN1 gene change, and a total systemic score of 7 or more together help doctors make the diagnosis."(Askari Nejad, & Aliakbar, 2025).A bulge or tear in the main heart artery: If the aorta size score is 2 or more in adults (3 or more in children), it strongly suggests Marfan Syndrome even if other signs are not present. (Sabahi Zadeh, 2025).This rule shows the serious blood vessel dangers of the syndrome and explains why checking the heart and blood vessels is very important for early detection.

5.2.1. Ectopia Lentis:

The lens of the eye is displaced upward and to the side, which is a very specific eye sign of Marfan Syndrome. "If it is present, especially together with a harmful FBN1 gene change, it is enough to make the diagnosis. (Frontiers in Cardiovascular Medicine, 2025). Lens subluxation happens in about 50 60% of patients and can come before heart problems, so eye check-ups are very important. (QJM, 2024).

5.2.2. Harmful FBN1 gene change:

Checking the genes is becoming more important. Finding a disease-causing FBN1 gene change through sequencing gives a clear diagnosis, even if bone problems are mild. More than 3,000 harmful gene changes have been reported, including different types like missense, nonsense, splice-site, intronic, and synonymous mutations, showing that Marfan Syndrome has many genetic differences (Yilmaz, Kaya, & Demir, 2024).Marfan syndrome influences the body by making connective tissue weaker, which normally holds organs and structures together. It commonly leads to a tall build with long arms, legs, fingers, and toes. The heart and blood vessels are often affected, mainly the aorta, which can stretch over time. Many people also experience eye problems and very flexible joints. With good medical care and routine checkups, most individuals with Marfan syndrome can live normal, active lives (Kaya, & Demir, 2024).

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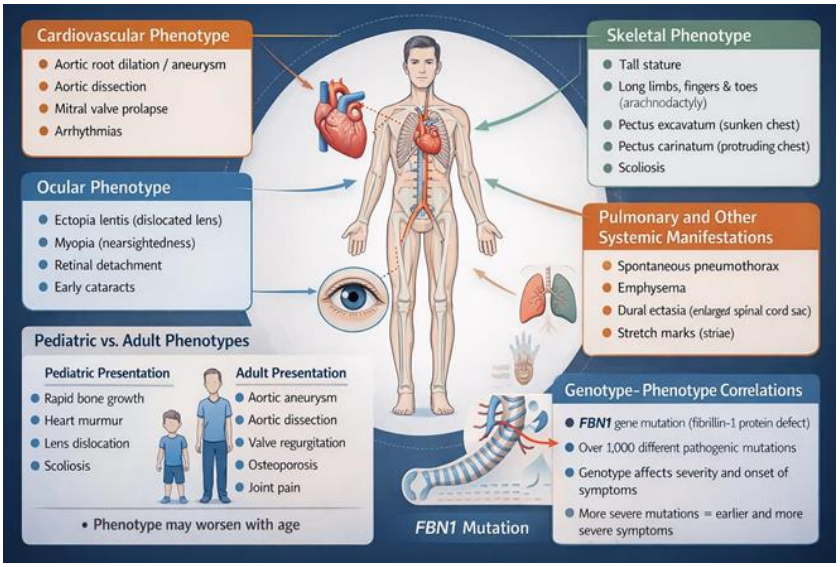

Fig. 03: Marfan syndrome Overview

5.2.3. Systemic Score: The systemic score measures body signs in different organs, including bones, skin, lungs, and face. A total score of 7 or more helps confirm the diagnosis and avoids wrong diagnosis in tall people whose signs look like Marfan syndrome but do not really affect the body systems. (Dove press, 2024).

5.3. Imaging Modalities

Scans are very important to find and watch blood vessel problems, which are the main causes of sickness and death in Marfan Syndrome (Rapezzi & Arbustini, 2023).

5.3.1. Echocardiography: The first-choice scan, which does not need surgery, is done once a year in teenagers and adults. It checks the size of the main heart artery and the ascending aorta, and finds mitral valve problems, giving an immediate look at heart and blood vessel risk. (Modrzyk et al., 2022).

5.3.2. CT Angiography: Provides clear scans to see bulges or tears in the aorta, especially in urgent situations. CT allows 3D images to help plan surgery, but it is not used much in young patients because of X-ray exposure. (Giannopoulos *et al* 2025)

5.3.3. MRI: A safe scan without radiation, considered the best choice for long-term check-ups. It is perfect for children, pregnant women, and for finding dural ectasia or deep blood vessel problems, working together with echocardiography and CT. (Rinaudo & Aurigemma, 2024).

5.4. Genetic Testing

FBN1 Sequencing: Gene testing using methods like Sanger sequencing, NGS, or MLPA can find harmful FBN1 gene changes. Finding a harmful FBN1 gene change not only helps confirm the diagnosis but also helps check family members and understand their risk. (Identification of two novel large deletions in FBN1 gene, 2024). De Novo Mutations: About 2530% of cases happen as new mutations, often showing more serious heart and blood vessel problems. Knowing about new gene changes is very important for correct diagnosis and advice in patients who have no family history of Marfan Syndrome (Ribeiro et al., 2023).

Some types of gene changes are linked to certain body features or symptoms. The kind of genetic change a person has can affect what physical traits appear in them. (BMC Pediatrics Early-Onset Marfan Syndrome Collaboration. (2023). Small protein-change types in certain protein parts are linked with serious problems in the main heart artery. Shortened gene changes raise the chance of the eye lens shifting from its normal position. Gene parts ,which often develop mutations, are linked with fast-worsening problems in the main artery. (Geiger et al 2023). Genotype phenotype spectrum and prognosis of early onset Marfan syndrome, 2023).

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5.5.Differential Diagnosis

Marfan Syndrome should be considered separated and not mixed with similar body support tissues conditions like Loeys–Dietz Syndrome (LDS) which is a genetic disorder fast spreading related to blood vessels, swollen blood tubules, head and face related abnormalities due to specific gene changes (Nistri et al., 2021). While Ehlers Danlos Syndrome is a connective-tissue disorder with extra stretchy skin, loose joints, and only a small amount of aorta involvement except in the blood-vessel type (Oldenburg et al 2024). Whereas, Homocystinuria is a metabolic disorder seen with a tall slim body shape, a lower-nose eye-lens that moves out of place, mental impairment, and blood clots (Loeys et al., 2021). Figuring out which disease is very important to help monitoring, treatment, and DNA advice, because the expected outcome and treatments vary a lot within these disorders (Loeys et al 2021).

6.Management and Treatment of Marfan Syndrome (MFS)

treatment and Management of Marfan Syndrome (MFS). Marfan Syndrome (MFS) management is a complex matter aimed at preventing life-threatening cardiovascular issues, observing systematic progression, and enhancing life quality with the help of specific interventions. MFS is a multi-systemic disorder that needs a multidisciplinary approach that involves cardiology, cardiovascular surgery, ophthalmology, orthopedics, genetics and where necessary pulmonology and neurology. (Lacro et al., 2014).

6.1.Cardiovascular Management

The most frequent causes of morbidity and mortality rates in MFS are cardiovascular complications, specifically, aortic root dilation and dissection (Milleron et al., 2023). The management strategies are supposed to decrease the stress on the aortic walls and slow down the dilation and rupture should it occur.

6.2.Pharmacologic Therapy

Pharmacologic treatment is the initial treatment to prevent aortic enlargement and to decrease the risk of dissection: Beta-blockers (e.g., at enol, propanol): Beta-blockers are considered the traditional therapy, which lowers heart rate and left ventricles ejection velocity, thus decreasing the stress of aortic walls (Milleron et al., 2023). Long-term beta-blockade has demonstrated to slow the aortic root enlargement rate particularly in children s populations. Angiotensin receptor blockers (ARBs, e.g., losartan, valsartan): ARBs block TGF-2 signaling that has been dysregulated in MFS and promotes the change in extracellular matrix and aortic wall degeneration (Cheng et al., 2023). Some randomized trials have shown that ARBs are capable of slowing or even halting aortic root growth either as an isolated intervention or with the usage of beta-blockers (Kim et al., 2023).

6.3 Surgical Interventions:

Surgical intervention is recommended in progressive dilation of the aortic root, valve dysfunction or complication like dissection. Modern practice suggests that prophylaxis may be performed in adults with a diameter of above 50 mm aortic root or less invasive thresholds (45 mm and below) in patients with high-risk characteristics like family history of dissection, rapid growth greater than 3mm/year (Milleron et al., 2023). Valve-sparing aortic root replacement (VSRR) Spares the native aortic valve in the patient and replaces the diseased root and eliminates the necessity of anticoagulation throughout life as well as enhancing long-term survival (Fukuda et al., 2022).

6.4 Ocular Management

Ophthalmologic evaluations: It is recommended that children and adults should have their eyes checked on a regular basis, either once in a year or once every two years to check the lens position, lens refractive errors, and early cataracts (Arbustini et al., 2014). Surgical lens correction: It is advised in severe cases of ectopia Lentis that result in visual impairment as it has to be meticulously planned to prevent retinal detachment or glaucoma.

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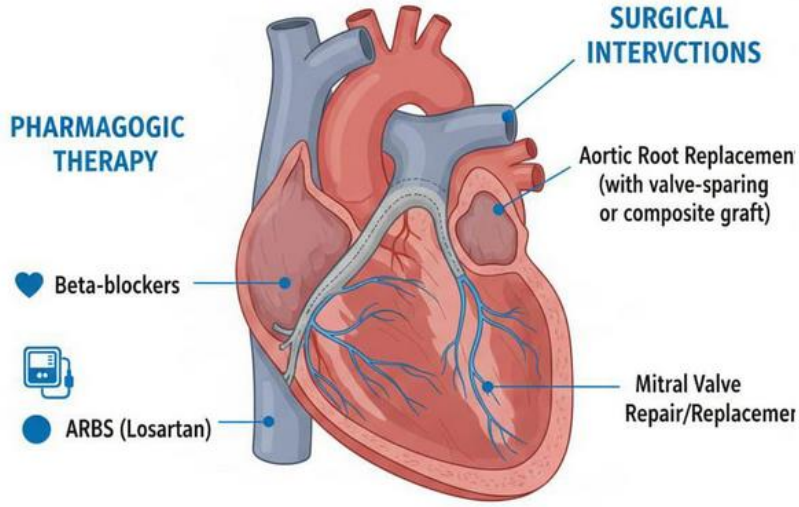

Fig. 04: Cardiovascular Management

6.5 Skeletal and Musculoskeletal Management

Skeletal appearances do not constitute life threatening conditions but can affect mobility, posture, and quality of life in general. Severe scoliosis, pectus abnormalities, or instability of the joints might require surgical treatment (Lacro et al., 2014).

Physical therapy: The focus is on the low-impact physical activities like swimming and cycling that help to maintain muscle tone without much strain on the aorta and joints. The absence of high-impact sports: Clients are recommended to avoid weight lifting and contacting sports, which may lead to cardiovascular risk. Pulmonary, Dermatologic, and other Systemic Management. These manifestations are rarer, but they have to be addressed specifically: Spontaneous pneumothorax: Conservative management or surgery, according to the severity; and smoking cessation is highly advised (Loeys et al., 2010). Sleep apnea and restrictive lung disease Thoracic deformities: In severe cases, it may necessitate lung functions monitoring and non-invasive ventilation procedures (Arbustini et al., 2014). Dural and skin appearances: Striae (stretch marks) are not typically problematic; dural ectasia can be treated or need a neurosurgical surgery when symptomatic (Radonic et al., 2011). New Molecular and Gene-based Therapies. Current studies are trying to address the molecular pathology: TGF-B regulation ARBs remain the gold standard; however, other anti-TGF-B options are actively examined to decrease the degeneration of the aortic wall (Cheng et al., 2023).

6.6 Gene therapy and CRISPR-based approaches:

The experimental models that fix FBN1 haploinsufficiency were found to reduce aortic dilation and enhance microfibril integrity, which is in the future perspective of personal therapy (Takeda et al., 2023). Stabilization agent of matrices: Clinical trials on elastin/ECM remodeling are aimed at reinforcing the vascular tissue and avoiding the formation of aneurysms (He et al., 2020). Genetic Counseling and Family Screening. Due to the autosomal dominant pattern of MFS, the family screening is essential: Genetic counseling gives education about the risk of inheritance, reproductive, and early detection (Rommel et al., 2020). Genetic testing in relatives based on cascade enables the pre-symptomatic tracking and prophylaxis in response to interventions to prevent morbidity and mortality (Takeda et al., 2023).

6.7 Lifestyle and Preventative Measures

This is lifestyle modification that augments medical management: Exercise in low impact that does not strain the aorta to keep the heart in shape regularly. Reduction of wall stress by blood pressure regulation that is usually accompanied by a medication. Prevention of smoking, stimulant and competitive contact sports. Early detection of any complication in new cardiovascular, ocular or skeletal symptoms. The prognosis and lifespan in Marfan Syndrome Prognosis. Interpretation: Diagnosis in early stages, pharmacologic treatment as well as elective surgery on time are life-

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saving. Life-long follow-ups are necessary to long life expectancy, MFS patients can have chronic pain, reduced physical activity, visual loss and cardiovascular risk anxiety. Genetic counseling, mental health, and psychological counseling Multidisciplinary support, such as social support, is critical to ensure functional independence and mental health (Radke et al., 2020). Case Studies and In-Real World Results in Marfan Syndrome. Case studies are helpful in creating practical evidence of clinical variability, diagnostic difficulties, and outcomes of Marfan Syndrome (MFS). They show the effects of early diagnosis, timely interventions and multidisciplinary care on prognosis.

7. Conclusion

Marfan Syndrome is a severe genetic condition of the body s support tissues that is uncommon and can happen because of harmful changes in the FBN1 gene. Its signs are many different and it affects the heart and blood vessels, eyes, bones, lungs, and skin. . Thats why finding it early and doing a full check is very important. This problem is treated by a team of doctors. The most important part of care is protecting the heart and blood vessels, which can be done with medicines (beta-blockers, ARBs), planned aortic surgery, and regular checkups for life. Patients show problems in bones, eyes, and lungs, which need special care with bone treatment, eye care, and lung monitoring to make sure the body works properly and life is comfortable. Patients show problems in bones, eyes, and lungs, which need special care with bone treatment, eye care, and lung monitoring to make sure the body works properly and life is comfortable.

Recent cell-level treatments and gene-based therapies, like TGF-2 medicines and CRISPR methods, look hopeful in changing the disease in the future. Using better ways to find the disease early, personal medical treatment, and quick surgery, people with Marfan Syndrome can now live nearly like normal people Genetic, medical, and scan data can be combined to allow a more personal check-up and management of the risk, which will later improve living longer and life comfort. In summary, Marfan Syndrome is not now a disease for life. By careful check, team care, and the chance of new cell-level treatments, people with the disease can live a long, healthy life and also lower the risk of serious problems.

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