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AN INTERESTING RARE CASE OF PROGERIA

General Medicine

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ABSTRACT

Introduction: Progeria is an extremely rare genetic disorder characterized by accelerated aging in children. It is caused by a de novo point mutation in the LMNA gene, leading to the production of an abnormal protein called progerin, which disrupts cellular function and integrity. The condition presents with distinct clinical features and significant cardiovascular complications, resulting in a markedly reduced life expectancy. **Case Report:** We present the case of a 17-year-old female with probable Progeria who exhibited a history of irregular menstruation (menorrhagia), shortness of breath on exertion, generalized weakness, and episodes of dizziness over the past 6 to 12 months. The patient displayed classical phenotypic features including, alopecia, a prominent forehead, a beaked nose, and thin lips. Despite these physical manifestations, the patient's intellectual and skeletal development remained normal. The diagnosis of HGPS was not confirmed through genetic testing. **Conclusion:** This case emphasizes the importance of early recognition and diagnosis of Progeria with unusual presentation and monitoring for life threatening Complications to optimize clinical management and improve the quality of life for affected individuals. As Progeria remains a rare and under-recognized condition, further research is essential to explore its pathophysiology and Develop more effective therapeutic strategies.To reassure patient and all Affected parties and bring them in confidence for counselling regarding family planning.

KEYWORDS

Progeria, LMNA mutation, accelerated aging, cardiovascular complication

INTRODUCTION

Progeria, is a rare genetic disorder characterized by rapid aging in children. It affects approximately 1 in 4 million live births. Children with Progeria exhibit significant growth retardation, alopecia, and distinct facial features such as a prominent forehead, beaked nose, and thin lips. Cardiovascular complications, including atherosclerosis and myocardial infarction, are common and contribute to a shortened life expectancy. The disorder is caused by a de novo point mutation in the LMNA gene, leading to the production of an abnormal lamin A protein called progerin. Progerin disrupts the nuclear envelope and cellular functions, accelerating cellular aging. Despite the physical symptoms, intellectual development and neurological function are usually normal. Early diagnosis is crucial for managing the disease effectively. Management involves a multidisciplinary approach, including growth hormone therapy and cardiovascular monitoring. Regular follow-ups help address various complications. The rarity of Progeria can lead to underrecognition, highlighting the importance of awareness among healthcare professionals. Current treatments focus on alleviating symptoms rather than curing the disease.

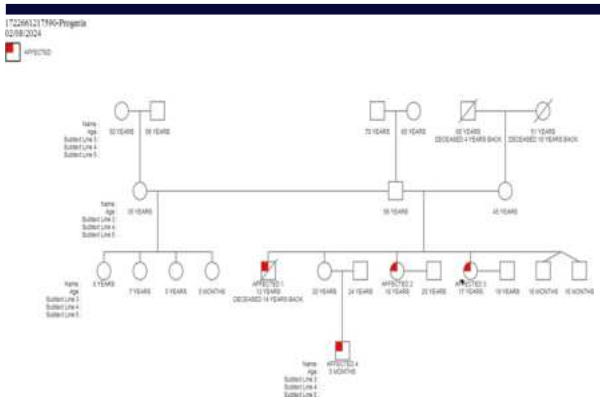
Ongoing research aims to better understand the condition's pathophysiology and develop new treatments. Experimental therapies, such as farnesyltransferase inhibitors, are being investigated. These therapies target the defective protein processing in Progeria. Genetic counseling is recommended for families of affected individuals. While life expectancy is significantly reduced, advances in care have improved quality of life. Support groups and specialized care centers provide essential resources for families. Research continues to seek a cure and improve therapeutic options. Early intervention and comprehensive care are key to managing Progeria's impacts.

Case Report

A 17-year-old female presented with a history of irregular menstruation (Menorrhegia), Shortness of breath on exertion, generalized weakness and episodes of dizziness since last 6 to 12 months. She exhibited the classical phenotypic characteristics of Progeria .Height and weight above the 3rd percentile, not consistent with Progeria growth patterns. She presented with Greying of hairs, alopecia, and features of premature aging of skin with Craniofacial Features of Micrognathia, beaked nose, prominent eyes .No significant clinical cardiovascular abnormality or history is present .No significant cognitive impairment or skeletal involvement noted. Normal mental development for her age.

On taking family history She has 1 sister,1 nephew and 1 brother affected with same clinical abnormality based on that pedigree is prepared as below.

Affected 3



Pedigree Chart

Affected 1

Affected 2

Affected 4

Lab Investigations

All routine investigations are normal. S.prolactin-43.7 ng/ml (<20ng/ml)

Haematological investigations suggestive of iron deficiency anaemia with microcytic hypochromic picture and normal liver function, lipid profile and kidney function test.

Radiological Investigations

Chest x-ray PA view had no skeletal or lung parenchymal abnormality. Skeletal survey x-rays are normal.

Ultrasound abdomen and pelvis were suggestive Poly cystic Ovarian disease.

CONCLUSION

This is a rare of rarest case of 17 year female with phenotypic features of Progeria , presented with irregular menstruation and anemia with no cardiovascular or neurological complications .This is unusual presentation for probable progeria which requires further genetic mutation testing for patient and her family members to confirm diagnosis and monitoring for any cardiovascular ,neurological or skeletal complications and Provide psychological support.

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