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Case report

A CASE OF PROGERIA SYNDROME TREATED AS VIP PATIENT

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ABSTRACT

Progeria is rare autosomal recessive genetic disease with an incidence of about one in eight million. He was 16 years old boy lying on the couch. He was short stature thin with minimal subcutaneous tissue, skin was thin and fragile with loss of hair over scalp, eyebrows and eyelashes, and his face was dismorphic with prominent eyes, beaked nose, small jaw and large cranium with visible veins over it. His voice was thin and high pitched. Overall, this gives them an extremely aged nearly 70 -80 years old man look. The patient was a known case of progeria syndrome and he was treated as a VIP patient by all faculty members and staff, though he belongs low socioeconomic status, no political issue with them. But still he was a VIP.

Keywords: Progeria, VIP patient, old man look (accelerated aging).

INTRODUCTION

Progeria is rare autosomal recessive genetic disease with an incidence of about one in eight million.[1] Prominent characteristic feature is accelerated aging. The classic type of progeria, is Hutchinson-Gilford Progeria Syndrome (HGPS), named for the two doctors who first described it.[2] The cause of Progeria is a mutation in the gene that encodes the protein Lamin-A.[3] Clinical presentation usually slow growth, slow weight gain, failure to thrive, with poor, very slow weight gain over time, loss of body fat and hair, aged-looking skin in spite of no abnormalities of growth hormone, parathyroid hormone, and adrenal hormones. Most of the patients die from complications of atherosclerosis, such as heart attack or stroke.[4] No definitive therapy is available.[2]

CASE REPORT

I am Dr. Seema mahant want to share a very interesting case experience. One day when I entered in the ward, I saw everyone was excited and running in one direction, I thought some VIP patient get admitted that's by everyone was thrilled. Eagerly I also went in the same direction. When I saw the patient I get surprised, he was 16 years old boy lying on the couch. He was short stature thin with minimal subcutaneous tissue. His skin was thin and fragile with loss of hair over scalp, eyebrows and eyelashes. His face was dismorphic with prominent eyes, beaked nose, small jaw and large cranium with visible veins over it. When I asked his name, his voice was thin and high pitched. Overall, this gives them an extremely aged nearly 70 -80 years old man look. He was very kind and cooperative. For this man his life is very fast days are becoming months, months become years and years became decades. He was a school going boy studying in 7th class and good in study. He is very friendly. On asking the complain patient told he have a cough and fever for 5 days. On

examination-patient was conscious oriented and well cooperative, he has mild fever, pulse 92 bpm, BP was 170/100 mmHg, respiratory rate was 30 per min. with minimal crepitition on Rt. lower chest.

Biochemical investigations were normal except for increased serum cholesterol. X-ray chest shows Rt. Lower pneumonitis with mild cardiomegaly. ECG shows-LVH with poor progression of R wave V1-V6. Echocardiography reveals- LVH with Gr. II diastolic dysfunction and poor LV function LVEF-40%. Treatment started with antibiotics, antihypertensive, lipid lowering drugs, antiplatelets and antianginal drugs. The patient was discharged after 7 days.

DISCUSSION

Progeria is a debilitating, rare illness anda genetic disorderr with just 45 odd cases in the world and is characterized by features of premature aging.[1] The probable cause is a mutation in the Lamin located in the nuclear matrix. An increase in the blood hyaluronic acid levels is responsible for sclerodermatous changes and cardiovascular abnormalities[2.] The classic type of progeria is Hutchinson-Gilford Progeria Syndrome (HGPS), named for the two doctors. Specific features of HGPS include- progressive heart disease, severe failure to thrive, with poor, very slow weight gain, over time, loss of body fat and hair aged-looking skin hearing loss stiff, painful joints, especially in the hips and feet, limiting range of motion hip dislocation bone rigidity and loss of bone mineral density, dry eyes, sometimes leading to irritation and clouding dental and gum disease.[3] Radiography of the skull shows craniofacial disproportion, delayed and abnormal dentition. Urine test results excessive excretion of the glycos aminoglycan, hyaluronic acid.[5]

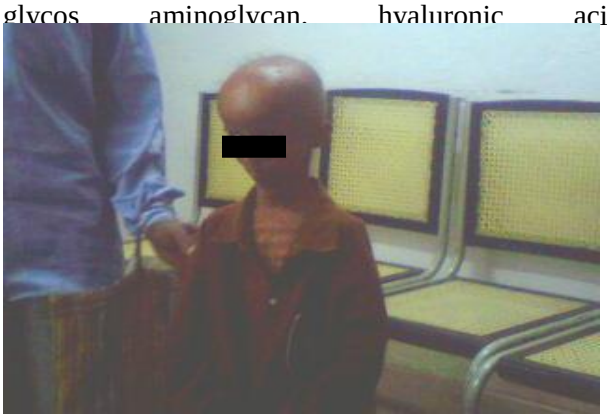

Fig 1: Slowed growth, with below-average height and weight, accelerated aging, prematurely old.

In this syndrome, the rate of ageing is accelerated up to seven times that of normal. The average life span is 13 years (range 7-27 year). The death is mainly due to cardiovascular complications like myocardial infarction or congestive heart failure. Till date, no definitive therapy is available and the patient is generally treated conservatively. [4, 5]

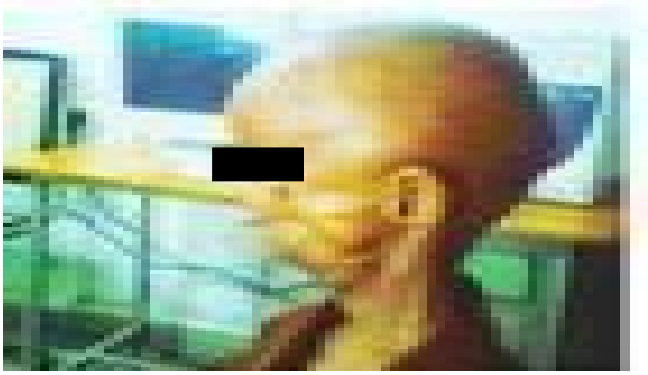

Fig 2: Showing Narrowed face, small lower jaw, thin lips and beaked nose, Head dis- proportionately large for face, Prominent eyes and incomplete closure of the eyelids, Hair loss, including eyelashes and eyebrows, thinning, spotty, wrinkled skin, Visible veins.

CONCLUSION

In our clinical practice, we see many patients daily, but few of them we cannot forget. Here I shared an interesting presentation of a rare disease Progeria. Right now the patient is not with us, but his memories will remain forever**.**

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Conflict of interest-: Nil

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