Case Report

Progeria syndrome: A rare combination of premature aging and dwarfism

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Abstract

We report a rare case of Progeria syndrome in 11 year old male child presenting with short stature, full body alopecia and striking appearance (small face with shallow recessed jaw, beak nose, prominent eyes, wrinkled skin with prominent scalp veins).Imaging studies supports the diagnosis of Progeria syndrome. It is rare genetic disorder with premature aging and dwarfism. Clinical and radiological feature mostly helps in diagnosis and confirmed by genetic test.

Keywords: Progeria, syndrome, Premature aging, Dwarfism.

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INTRODUCTION

Progeria syndrome is extremely rare genetic syndrome that cause premature aging shortly after birth .The classic form is known as hutchison gilford syndrome. It occur sporadically with reported incidence of 1 in 4 to 8 million with M: F ratio of 1.5:1. Their average life span is 13 years (range 8-21) and death is mainly due to cardiovascular complication. Progeria is rare combination of dwarfism and premature aging. It is a genetic condition that occur as a new mutation and is rarely inherited[1,2,3]. Though children with progeria are born healthy, they usually develop their first symptoms during their first few months which include failure to thrive and localized scleroderma like skin condition and post infancy present with limited growth, full-body alopecia, and a striking fascies (small face with beak nose with shallow recessed jaw)[4]. we report such a rare case in 11 years old male child.

CASE REPORT

11 years old male child presents to us with short stature, abnormal fascies and coarsening of skin. He was product of non consanguineous marriage without any family history of major medical or genetic disorder. He was born normally at term with birth weight of 2.5 kg. The perinatal history was uneventful. He was apparently normal till age of 1 year when their parents notice features like coarsening of skin, global alopecia and retardation of growth. O/E child was short stature and malnourished with anthropometric measurement of height 98 cm, weight 9 kg, head circumference 44 cm, chest circumference 42 cm and US/LS ratio of 1.45 . He had striking fascies (Fig 1a and 1b) with prominent eyes, beak nose sparse eyebrows and eyelashes, micrognathia, global alopecia and prominent scalp veins, sclerodermatous change over extremities, prominent elbow joints, difficulty in changing posture, dystrophic nails and clawing of hands .He had normal intelligence. Complete haemogram, erythrocyte sedimentation rate, routine urine investigations, liver and renal function tests, blood sugar levels and electrocardiogram studies were within normal limits. Opthalmic examination did not reveal any abnormalities. Imaging studies were as follow: X-ray Skull (Fig 2) shows multiple wormian bones, increase diploic space and diastasis of sagittal suture, with hypoplastic mandible. X-ray chest(Fig 3) shows absence of the lateral half of clavicle and thin ribbon like third rib on both sides .X-ray pelvis and long bones (Fig 4 and Fig 5) shows bilateral coxa valga deformity, shortened long bones with constricted central segments and demonstrate flares at the ends. X-ray hands and feet **(Fig 6)** shows resorbtion of phalanges and acro-osteolysis.

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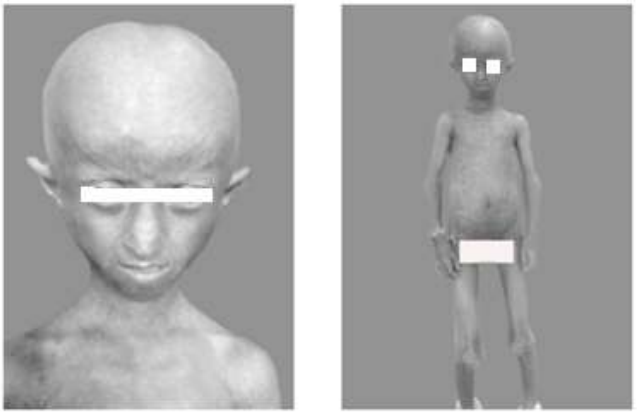

Figure 1a and 1b: The features of progeria syndrome in our patient

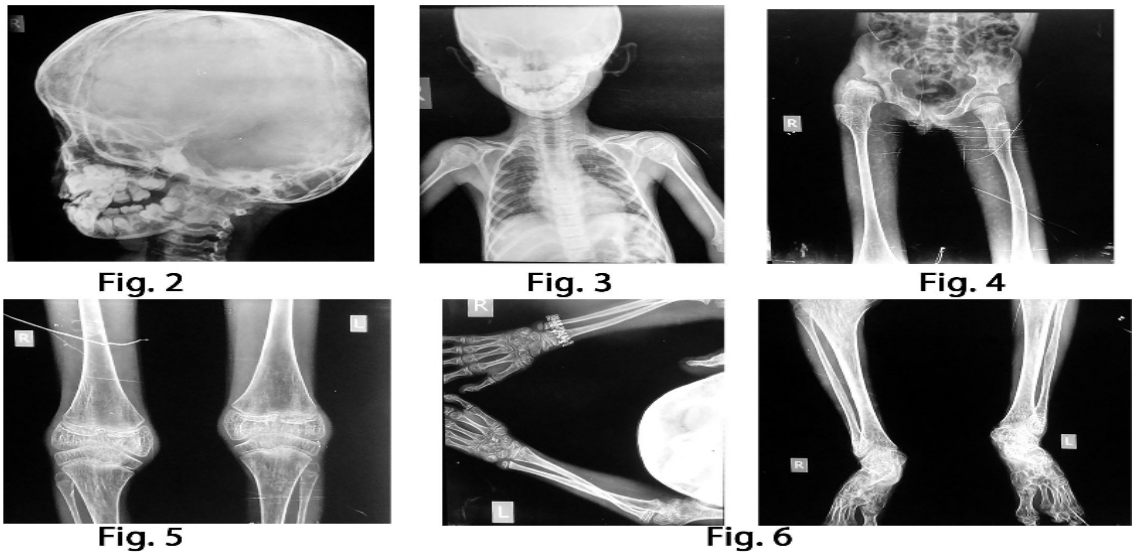

Figure 2,3,4,5,6: Radiological features of progeria syndrome in our patient

DISCUSSION

Progeria syndrome is an extremely rare genetic disease wherein symptoms resembling aspects of aging are manifested at a very early age. The disorder has a very low incident rate, occuring in an estimated 1 per 4 to 8 million live birth. It is genetic condition that occurs as a new mutation and is rarely inherited. Children with progeria are born looking healthy, when they are about 10-24 months features of accelerated aging starts to appear[1,2,3]. Children who suffer from progeria are genetically susceptible to premature and progressive heart disease and death is mainly due to cardiovascular complication like myocardial infarction or congestive heart failure[3,5]. It is believed that progeria is caused due to LMNA (lamin A protein) gene mutation on chromosome[1], resulting in defective lamin A protein called progerin. Normal lamin A protein holds the nucleus of cell together while defective lamin A protein (progerin) make nucleus unstable. This instability seems to lead to process of premature aging among progeria patients[6,7]. Affected children usually have growth failure, loss of hair, loss of body fats, wrinkling of skins, stiffness in joints, cardiovascular disease like hypertension, stroke, angina ,enlarged heart,and heart failure. The differential diagnosis includes Werner syndrome, Acrogeria, Rothmund-thompson syndrome, Cockayne syndrome. But typical phenotypic and radiological features differentiate progeria from other as in our case. A genetic test for LMNA mutation can confirms the diagnosis of progeria[2,8]. Till date there is no cure for progeria syndrome. Associated arthritic, respiratory and cardiovascular problem are mainly treated symptomatically. But recent clinical drug trial treatment with Farnesyl transferase inhibitor currently used for treating cancer have shown significant improvement in bone structure, weight gain and cardiovascular system[9]. Our case have striking clinical and radiological feature of progeria syndrome. As facility of genetic analysis is not

available in our institution, we have refer patient to higher centre. Detail research on progeria syndrome can advance our understanding of process of premature aging and atherosclerosis which may provide the key for youthness. But are we really ready for that?

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