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A Rare Case Report: Hutchinson-Gilford Progeria Syndrome

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ABSTRACT:

Hutchinson- Gilford progeria syndrome is a very rare genetic disorder in children which causes accelerated aging involving bones, heart, skin and blood vessels. This condition is extremely rare, affecting only 1 in 4-8 million births.The features include premature aging, alopecia, cardiovascular problems, sclerodermatous skin changes and bone fragility. The main cause of death is Myocardial infarction, usually happens at an average age of 13 years. We are reporting a 2 year old boy showing features of this syndrome who was proven genetically as well.

Keywords: Progeria, Hutchinson - Gilford syndrome, accelerated cellular aging.

1. Introduction

HGPS is an extremely rare and debilitating genetic disorder that affects approximately 1 out of 4 million live births [1]. First described by Jonathan Hutchinson and by Hastings Gilford in 1897[2]. It is characterised by accelerated aging in children, leading to range of devastating physical symptoms that resemble premature aging.

Also known as Progeria, this syndrome occurs due to a point mutation in the LMNA gene, encodes for Laminin A protein essential for nuclear stability [3]. This mutation leads to a protein which is toxic that causes accelerated cellular aging, resulting in symptoms that progress rapidly[4].These patients have a characteristic "plucked-bird" appearance[5].

Despite their physical limitations, children with HGPS typically have normal cognitive development and intelligence [6]. With the median life expectancy of 13-20 years, HGPS is a fatal condition that requires urgent medical attention, genetic counseling, supportive care to manage its symptoms [7].

Case Report

A 2 year old boy was brought to our Paediatric outpatient department with complaints of progressive loss of scalp hair,eyebrows and eyelashes since 6 months of age. Antenatal history was normal for this child and was born to non-consanguineous parents.

On general and physical examination, the child had a distinctive appearance with plucked-bird look, bulging eyes,visible scalp veins, loss of hair over the scalp, eyelashes, eyebrows and a senile look(figure1,2). There was mottled pigmentation of skin over the lower limbs and trunk (figure 3). Clinical evaluation revealed joint stiffness, hands were short with hardening and thickening of skin over knuckles (figure 4). The ribs were prominent. The lower limbs also had prominent knees and there was a slight leg deformity. The child had normal cognitive development and intelligence. Based on these findings, we suspect the child to be having Progeria.

Routine investigations were normal except a slightly low value of high density lipoprotein (HDL). To confirm the diagnosis, radiological investigations were done. 2D echo -Normal. Karyotyping- normal. Genetic study revealed LMNA -exon 11 mutation which confirmed the diagnosis of Progeria syndrome.

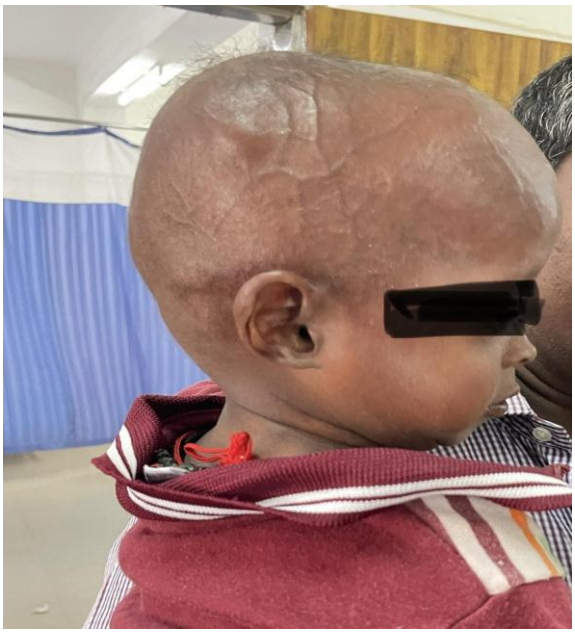

Figure: 1 Prominent scalp veins with sparse hair.

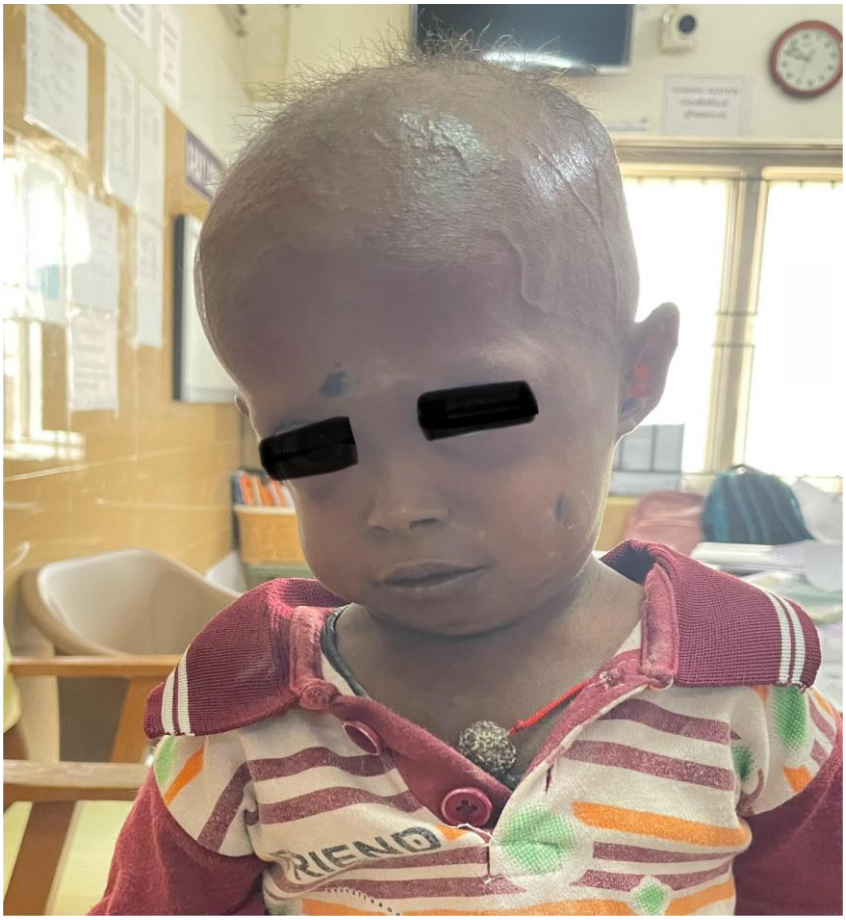

Figure : 2 The typical face: senile look, bulging eyes, thin hair, receding chin, pointed nose.

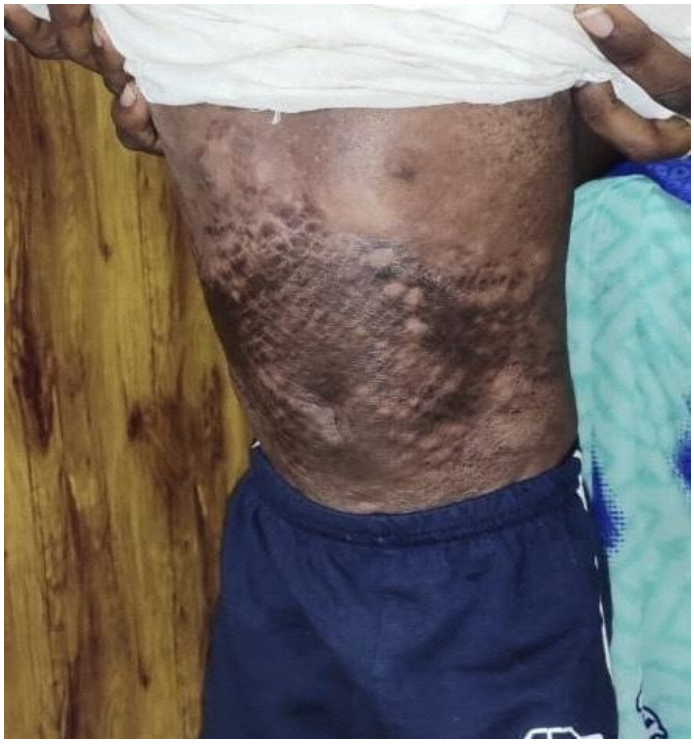

Figure: 3 Mottled pigmentation of skin with changes over the trunk.

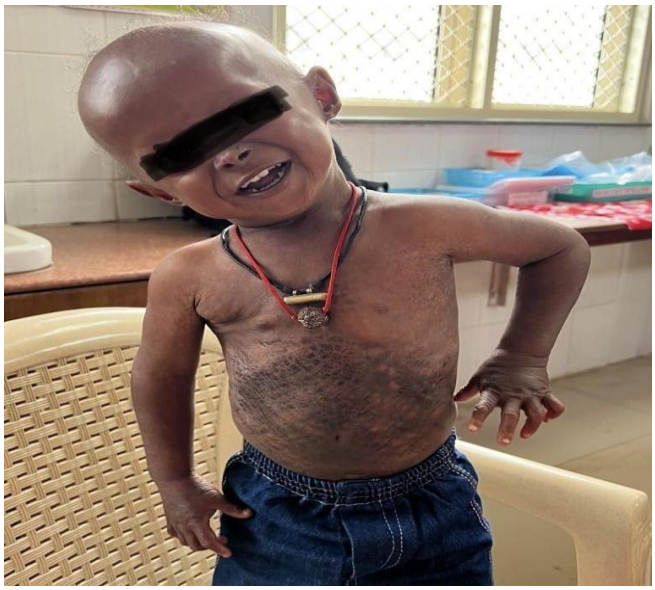

Figure: 4. Child with unusually small hands and clawed fingers with skin over knuckles abnormally thick and hardened.

2. Discussion

Most cases of the Hutchinson-Gilford progeria syndrome occur due to autosomal dominant mutation of the LMNA gene. This mutation leads to abnormal cell growth and division, damaged nuclear membrane, disrupted DNA repair and gene expression, shortened telomeres, cells aging and dying faster than usual [8].
The clinical manifestations include:
-Head appears abnormally large compared to face.
-Delay in eruption of teeth and crowding of teeth because of small mouth.
-Sparse hair with loss of eyebrows and eyelashes.
-Musculoskeletal defects like hip dislocation, osteonecrosis, with progressive stiffness of joints. -Cardiovascular problems developing atherosclerosis [9]
-Nocturnal lagophthalmos, corneal ulceration occurs due to exposure keratitis [10]
-Hearing defects mostly conductive hearing loss [11].
The child appears to be healthy at birth but may have mottled changes in the skin which also involves the trunk and also extremities in few cases [12]. In most of the cases, the changes appear within 1-2 years.
This case report highlights the rare condition of Progeria Syndrome, which causes accelerated aging in children. Our patient, a young child, exhibited typical symptoms such as sclerodermatous skin changes over trunk, hair loss,prominent eyes and wrinkled skin, despite having normal cognitive development.
This case report emphasises on the importance of early diagnosis and to start the multidisciplinary treatment. It also highlights the need for further research into the causes and treatment of Progeria, to improve the lives of affected children and their families.

3. Conclusion

This case report serves as a reminder of the importance of awareness and understanding of rare genetic disorders like Progeria, and the need for continued research and support for affected

families.Unfortunately, there is no cure, and management focuses on alleviating symptoms and supporting the child's quality of life. Certain therapies may delay or ease symptoms of progeria. They include Lonafarnib -this medicine prevents the buildup of faulty progeria and progeria like proteins in cells which slows the progression. Others include physical and occupational therapy, nutrition and hearing aids.

Consent: Obtained From The Guardian.

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