

Hutchinson-Gilford progeria syndrome: a rare case report

Kalegowda Deepadarshan¹, Bugude Gangadhar², Mallaiah Mallikarjun²

¹Department of Dermatology, Mandya Institute of Medical Sciences, Mandya-571401, Karnataka, India, ²Department of Dermatology, KVG Medical College and Hospital, Sullia, DK.-574327, India

Corresponding author: Dr. Kalegowda Deepadarshan, E-mail: deepadarshan.rani@gmail.com

ABSTRACT

Progeroid syndromes are characterised by clinical features of physiological aging at an early age. Hutchinson-Gilford progeria syndrome is a type of progeroid syndrome, characterised by abnormal facies, bone abnormalities, sclerodermatous skin changes and retarded physical development. Average life expectancy of progeria patients is 13 years. Herein we are reporting a case of progeria who is 21 years old.

Key words: Hutchinson-Gilford progeria syndrome; Short stature, Scleroderma

Introduction

Hutchinson-Gilford progeria syndrome is a rare hereditary disorder caused by mutations in lamin A gene which encodes proteins of nuclear lamina, results in production of a truncated protein called progerin [1]. This disorder was first reported by Hutchinson in the year 1886 [2]. Gilford introduced the term progeria. This syndrome along with Werner syndrome are also known as segmental aging syndromes, as they do not feature all aspects associated to physiological aging. Till now less than 100 cases of progeria have been reported in world literature [3].

Case report

A 21 year old male presented with pigmentation of face with itching as major complaint since 6 months. No history of photosensitivity. There was history of delayed physical development and dentition noted at 2yrs of age with no history of convulsions. Intelligence was normal. On examination he had typical bird like facies characterised by large cranium with frontal bossing, prominent eyes, beaked nose, prominent ears, sunken cheeks, micrognathia and mottled pigmentation (Fig. 1). In our patient, unlike progeria there was no loss of eyebrows and scalp hair.



Figure 1: Typical bird like facies with frontal bossing, prominent eyes, & micrognathia.

Detailed examination revealed short stature with height measuring 94 cm (Fig. 2). Nails were thin and brittle. Sclerodermatous thickening of skin noted over the dorsa of hands and fingers. Prominent joints, loss of subcutaneous fat and muscle atrophy were other important features noted. Ocular examination was within normal limit. Systemic examination were within normal limits. Radiological examination of skull, chest and extremities were within normal limits. Electrocardiography showed features of left ventricular failure.

How to cite this article: Deepadarshan K, Gangadhar B, Mallikarjun M. Hutchinson-Gilford progeria syndrome: a rare case report. Our Dermatol Online. 2016;7(2):210-212.

Submission: 01.08.2015; **Acceptance:** 04.10.2015

DOI: 10.7241/ourd.20162.58



Figure 2: Short stature of the patient.

Discussion

Hutchinson-Gilford progeria syndrome (HGPS) is a rare disorder, has an incidence of 1 in 4 to 8 billion newborns [4]. It occurs due to sporadic mutation, hence not run in families. Characterised by accelerated aging, short stature, alopecia, generalised atrophy of skin and muscles with sclerodermatous changes over the extremities. The facial appearance is reminiscent of a fledgling bird, with a disproportionately large cranium with patent fontanelles and frontal bossing, prominent eyes and scalp veins, very sparse, downy scalp hair, sparse or absent eyebrows and eyelashes, centropalpebral cyanosis, micrognathia, thin lips and a 'beaked' nose [5,6]. Bird like facies is not characteristic of HGPS, also seen in association with Hallermann-Streiff syndrome, familial partial lipodystrophy, Nijmegen breakage syndrome [7].

Progressive mottled hyperpigmentation of skin is noted over exposed parts with aging, but there is no photosensitivity. Abnormal and delayed dentition is noted and associated skeletal abnormalities such as dystrophic clavicles and coxa valga, with joint contractures and a 'horse-riding' stance also observed.

It has to be differentiated from other aging syndrome like Werner's syndrome (adult progeria) which usually

presents between 14 and 18 years of age with short stature, immature sexual development, cataract, glaucoma, and sclerodermatous changes, Cockayne's syndrome manifested as facial erythema in butterfly distribution, photosensitivity, ocular defects, and a "micky mouse" appearance with disproportionately large hands and feet and protruding ears, Rothmund-thomson syndrome appears between 3 and 6 months of age with poikilodermatous skin, photosensitivity, and cataract.

Our patient had retarded growth and accelerated aging since 2yrs of age, with typical facial features except for presence of eyebrows and scalp hair, short stature, sclerodermatous changes in skin, loss of subcutaneous fat and atrophy of skin and muscles, radiological features of osteopenia with features of left ventricular failure in electrocardiogram.

Complications include atherosclerosis, angina attacks, cerebrovascular accidents, osteoporosis, pathological fractures and dental anomalies.

Treatment is mainly symptomatic. Long term follow up of patient is required to observe skeletal and cardiovascular changes. Main aim of treatment is to control diabetes, osteoporosis and atherosclerotic changes. Low dose aspirin is used prophylactically to control ongoing atherosclerosis. Pravastatin and bisphosphonates like zoledronic acid to prevent atherosclerosis and osteoporosis is under trial. A phase II trial of lonafarnib, a farnesyl transferase inhibitor, in progeria is currently ongoing in USA [4].

Conclusion

Hutchinson-Gilford progeria syndrome is very rare disorder, less than 100 cases have been reported worldwide. Only few cases have been reported to survive after second decade. Because of its rarity, lack of reporting and long term follow up prompted us to report this rare syndrome.

Consent

The examination of the patient was conducted according to the Declaration of Helsinki principles.

REFERENCES

1. Eriksson M, Brown WT, Gordon LB, Glynn MW, Singer J, Scott L, *et al*. Recurrent de novo point mutation in lamin A cause Hutchinson-Gilford progeria syndrome. *Nature*. 2003;423:293-8.

2. Crounse RG. Clinical Dermatology Vol. I, Demis DJ. Sixteenth Revision, Philadelphia, J.B. Lippincott Company 1989, Unit 4 - 30, 1-5.
3. Badame AJ. Review of progeria. Arch Dermatol. 1989;125:540-4.
4. Agarwal US, Sitaraman S, Mehta S, Panse G. Hutchinson-Gilford progeria syndrome. Indian J Dermatol Venereol Leprol. 2010;76:591.
5. Hamer L, Kaplan F, Fallon M. The musculoskeletal manifestations of progeria: A literature review. Orthopedics. 1988;11:763-9.
6. Ganesh SP, Narendra KK, Maria K. Disorders of connective tissue. In: Valia RG, Valia AR, editors. IADVL Textbook of Dermatology, 3rd ed. Mumbai: Bhalani Publishing House; 2008. p.1181-3.
7. Kanathur S, Sarvajnyamurthy S, Somaiah SA. Characteristic facies: An index of the disease. Indian J Dermatol Venereol Leprol. 2013;79:439-43.

Copyright by Kalegowda Deepadarshan, et al. This is an open access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Source of Support: Nil, **Conflict of Interest:** None declared.