Journal of Optometry and Ophthalmology

Wadhwani M, 2020-J Opto Ophth
Case Report

Hutchinson Gilford Progeria Syndrome with Ocular Manifestation

Meenakshi Wadhwani*

Assistant Professor, Department of Ophthalmology, Chacha Nehru Bal Chikitsalya, New Delhi, India

***Corresponding Author:** Wadhwani M, Assistant Professor, Department of Ophthalmology, Chacha Nehru Bal Chikitsalya, New Delhi, India

Accepted Date: 11-04-2020
Published Date: 12-04-2020

Abstract

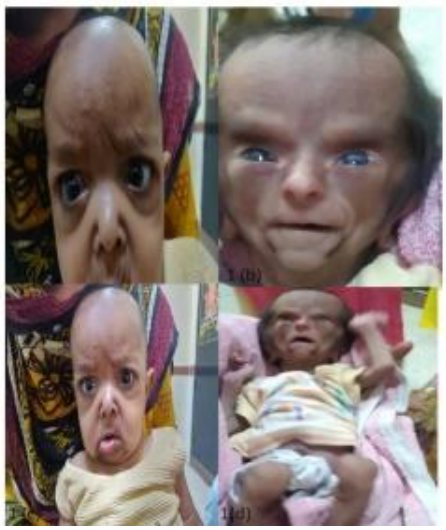
Hutchinson Gilford progeria Syndrome (HPGS) was first described by Jonathan Hutchinson Gilford in 1897 [1]. It is characterized by characteristic facies also described as plucked bird appearance. These are reported to occur due to denovo autosomal dominant mutation in Laminin A(LMNA) gene present on 1q21.1-1q 21.3 and are rarely inherited [2]. This is to report a rare case series of two children presenting with Hutchinson Gilford progeria with ocular manifestation.

Keywords: Hutchinso Gilford Progeria Syndrome; Ocular Manifestation; Rare Case; Conjunctival Granuloma

Case Report

Child 1 was a 15 month old female baby who presented to the ophthalmology outpatient department of a tertiary care hospital with swelling in right eye, Ocular examination revealed conjunctival granuloma in right eye (OD), rest of the examination of both anterior and posterior segment was unremarkable (Figure 1a). Child 2 was a 26 month old male baby who presented to our ophthalmology outpatient department with complaints of opacity in both eyes, ocular examination revealed exposure keratopathy due to lid retraction (Figure 1b), rest of the examination of both anterior and posterior segment of this child was also was unremarkable. Both the children could fix and follow at torch light with history of consanguineous marriage in parents, both had similar x-ray features of premature aging as acroosteolysis of tarsalis with open anterior frontenalle and were already diagnosed as Hutchinson Gilford progeria in pediatric medicine outpatient department (Figure 1c and d).

Figure 1: (a) Conjunctival granuloma in right eye; **(b)**exposure keratopathy due to lid retraction; **(c,d)** gross progeria facies



Discussion

The infant presenting with HPGS have sclerodermatous skin changes especially in the area of trunk and extremities. Hair growth is decreased over scalp and other parts of body Lipodystrophy over the face leads to typical senile look, glyphic nose and "plucked bird appearance"[3,4]. There is involvement of lower limb with valgus deformity. There is a cardiovascular involvement leading to death due to atherosclerotic changes owing to vascular involvement in second decade of life. It is also characterized by the presence of micrognathia, thin limbs, macrocephaly, prominent scalp veins, patent anterior frontanelle, short stature. The index cases also had micrognathia, decreased scalp hair, open anterior frontenelle and short limbs.

Its main differential diagnosis is Rothmund Thomson syndrome, Cockaynesyndrome, Werners syndrome. Rothmund Thomson syndrome that was ruled out by erythema, poikiloderma and cataract. Cockayne syndrome had a similarity of ocular defect but also was ruled out by absence of photosensitivity, facial erythema. Werners syndrome is ruled out by manifestations at an earlier age [5].

Although there are around 116 cases reported in the world of this HGPS [4,6] but this is another interesting case for academic interest with ocular changes. Proper counselling of parents is important for the diagnosis of this condition, for proper follow-up of different systemic conditions associated with it.

References

1. Hutchinson J. Case of congenital absence of hair, with atrophic condition of the skin and its appendages, in a boy whose mother had been almost wholly bald from alopecia areata from the age of six. Lancet1886;1:923.
2. Pollex RL, Hegele RA. Hutchinson – Gilford progeria syndrome. Clin Genet 2004;66:375-81.
3. Gordon CM, Gordon LB, SnyderB D, Nazarian A, Quinn N, Huh S, et al. Hutchinson-Gilford progeria is a skeletal dysplasia. J Bone Miner Res 2011;26:1670-9.
4. Stehbens WE, Wakefield SJ, Gilbert-Barness E, Olson RE, Ackerman J. Histologicalandultrastructuralfeaturesofatherosclerosisinprogeria. CardiovascPathol 1999;8:29-39.
5. Merideth MA, Gordon LB, Clauss S, Sachdev V, Smith AC, Perry MB, et al. Phenotype and course of Hutchinson-Gilford progeria syndrome. N Engl J Med 2008;358:592-604.
6. Kashyap S, Shanker V, Sharma N. Hutchinson- Gilford progeria syndrome: A rare case report. Indian Dermatol Online J 2014;5:478-81.