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Meier-Gorlin Syndrome With Pappilloedema

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Abstract

Meier – Gorlin Syndrome (Ear- Absent Patella –Short Stature Syndrome) is a rare autosomal recessive condition, only about 30 cases have been reported in literature so far[1]. First described by Meier& Rothschild[2]. In the past, Meier Gorlin syndrome has been compared and contrasted with some forms of primodial dwarfism. It is characterized by a triad of Microtia, Absent patelle, Growth retardation.

Key Words: Meier Gorlin, Pappilloedema

Case Report:

A 6 Year old boy born to non consanguineous parents was brought to our hospital with complaints of failure to thrive. On examination he had the following characteristics. Microcephaly, craniosynostosis, microtia, low set ears, small mouth with full lips, micrognathia and a high arched palate, prominent veins over nose and temporal region, crowded teeth with a beaked nose (fig 1).

He had a cheerful and friendly personality his skeletal examination reveled absent patella (fig 2), slender long bones, hyper extensible joints, chest asymmetry, scoliosis, fifth finger clinodactyly He also had absent testis on right side and on left it was in the inguinal region, scrotum was well developed.

His anthropometric measurements were height -91 cms, weight-8.6 k against the 113cm ht & 20kg expected values respectively. His auditory function was normal. His ophthalmologic evaluation revealed established papilloedema. He had two siblings who are normal with no craniofacial anomalies. Both his parents were normal and no such history in the family[3].

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Discussion:

Meier-Gorlin syndrome is a rare autosomal recessive condition with characteristic craniofacial abnormalities like microcephaly, microtia, low set malformed ears, small mouth with full lips,micrognathia, high arched palate, prominent veins over nose and forehead, characteristically absent patella or hypoplastic patellae along with short stature is seen, hyperextensible joints, chest asymmetry, campodactyly, clinodactyly and in case of females incompelete breast development and normal menstruation

is seen, cryptorchidism, micropenis and in one rare case severe deafness with congenital labrynthine anomalies was also seen, therefore neuroradiograhic imaging and functional inner ear investigations are recommended in the diagnostic workup of this rather rare and specific disease.there is also a difference in facial characteristics between patients reported early in infancy and those described at later age, follow up is required [4] some recommend radiographic survey of the patellae in patients at older age to investigate the weight of absent or hypoplastic patellae in the diagnosis syndrome.

Thus this case of Meier-Gorlin syndrome with pappiloedema,a characteristic which has previously not been described as a feature of this syndrome, therefore requires a detailed ophthalmologic

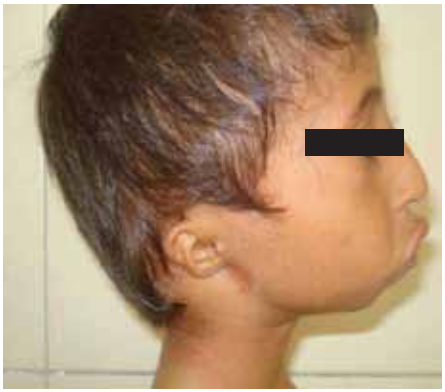

Fig. 1

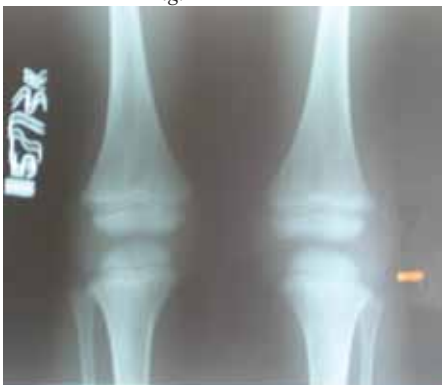

Fig. 2

evaluation, which is recommended in the diagnostic workup of this syndrome.

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