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CASE REPORT

Dentofacial characteristics in a child with Meier–Gorlin syndrome: A rare case report

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Abstract Meier–Gorlin syndrome (MGS) is a rare autosomal recessive disorder characterized by the triad of microtia, absent or small patellae and short stature. The other associated clinical features may include developmental delay, congenital pulmonary emphysema, gastro-esophageal reflux, urogenital anomalies, such as cryptorchidism and feeding problems. The facial characteristics during childhood are typical, comprising of a small mouth with full lips and micrognathia/retrognathia. The condition is rare affecting about one to nine individuals per million. Mutation in the genes of pre-replication complex involved in DNA-replication is detected in the majority of patients. This impedes the cellular proliferation resulting in a reduction of total cell number and thereby retardation of overall growth. This case report describe the typical dentofacial characteristics in a 5 years old child affected with Meier-Gorlin syndrome along with other associated anomalies and a multidisciplinary approach for their management.

1. Introduction

Meier–Gorlin syndrome (MGS) is a rare autosomal recessive disorder. It is characterized by the triad of short stature, absent or small patellae and microtia. In most of the patients with MGS at-least two of these three clinical features are present. The combination of patellar hypoplasia and microtia are most prevalent (De Munnik et al., 2012a). The patients can exhibit short stature, without microtia or patellar anomalies, indicating that the clinical phenotype can be variable (Bicknell et al., 2011).

The first case of MGS was reported by Meier in 1959 and Gorlin in 1975 and only 67 patients with MGS have been reported in the literature till date. There is no reported data on the prevalence of MGS, but it is estimated to be less than one to nine individuals per million. It is based on number of cases reported in the literature. Though, this might be an underestimated figure due to missed diagnosis and underreporting (De Munnik et al., 2015).

The underlying mechanism responsible for clinical features of MGS remains unknown. Mutations in five genes of the pre-replication complex have been detected in patients with MGS

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(De Munnik et al., 2012a). The impaired function of pre-replication complex is presumed to reduce the G1 phase of DNA replication. This limit the available time for origin licensing and retard the cellular proliferation resulting in a reduction of total cell number. This results in diminished overall growth (Klingseisen and Jackson, 2011; Kuo et al., 2012).

The diagnosis of MGS should be considered in a patient with short stature and microtia. The presence of these features necessitates a comprehensive examination of patellae. Ultrasound investigations are advised in children in the first five to six years of life since patellae are radiolucent and would not be visible in conventional radiography (De Munnik et al., 2015). Furthermore; the dentofacial characteristics are the specific findings for MGS and can assist in diagnosing the condition.

This paper describes a case of Meier-Gorlin syndrome with its dentofacial characteristics and suggests a multidisciplinary approach for its management.

2. Case report

A 5 year old child reported to the Department of Oral Health Sciences Center, PGIMER, Chandigarh with multiple carious teeth. Intraoral examination was suggestive of severe early childhood caries involving all primary teeth. The child had a small oral cavity with limited mouth opening. The family history was non-contributory with non-consanguineous marriage. No other family member was affected. The child had an overall short stature and the anthropometric variables like height, weight and occipitofrontal head circumference were deficient for age according to the WHO Child Growth Standards 2006 (Table 1). The prenatal history revealed intra-uterine growth retardation (IUGR) and microcephaly. The child was evaluated for endocrinal abnormalities and the results were within normal limit. Medical history was significant with a child suffering from recurrent seizures and was under oral phenobarbitone therapy for the same. The karyotyping results revealed 23 pairs of chromosomes (46, XY) without any abnormality or structural defects. The general examination was suggestive of developmental delay with delayed attainment of developmental milestones. There was overall growth retardation with delayed speech, short stature, and microcephaly. External ears were deformed indicative of mild microtia. A moderate hearing loss was present in both the ears. A reduced mouth opening with bilateral fibrous ankylosis of Temporomandibular joint was present. Other features included preaxial polydactyly at right hand, gastro-esophageal reflux and penile deviation. The renal ultrasound was suggestive of an ectopic kidney with normal functions. MRI brain revealed poor grey-white matter differentiation and partially deficient falx cerebri. It also showed absence of septum pellucidum with a few gyri interdigitating in the anterior hemispheric fissure (Table 2). The ultrasound examination of patellae was carried out as it is radiolucent in first 5–6 years of life and may not be visible by conventional radiography. The ultrasound revealed poorly developed patellar bone (De Munnik et al., 2015). The blood profile of was suggestive of anemic state with a normal coagulation profile.

Table 1 Anthropometric variables.

Anthropometric variables	Patient parameter (Deficiency)
Weight	7.4 kg (−7.49)
Height	79.6 cm (−5.86)
Occipitofrontal head circumference	39.6 cm (−9.24)

The facial examination was suggestive of a dysmorphic face with microstomia. Mandible was retrognathic with a prominent upper and middle third of face (Fig. 1a). The child had a reduced mouth opening since the birth. Intraoral examination revealed severe early childhood caries involving all maxillary and mandibular primary teeth (Fig. 1b).

Cleft involving the junction of hard and soft palate was present with an occasional nasal regurgitation. Cleft remained untreated due to reduced mouth opening. Radiographic examination with orthopantomogram (Fig. 2a) revealed a normal tooth development and the presence of developing permanent teeth expected for dental age. The cephalometric radiograph (Fig. 2b) confirmed the clinical observation of prominent upper and middle third of face with a retrognathic mandible. There was a deficiency in overall length of maxilla and mandible with an increase in the gonial and mandibular plane angle (Table 3).

The parents were explained about the complications that can occur after the birth. However, this disorder has a variable expression and relatively low chances of life-threatening complications or intellectual disability. As there were no structural congenital anomalies, the ethical issue of possible termination of a pregnancy was difficult to justify. MGS being an autosomal recessive disorder with a recurrence risk for a couple with an affected child is about 25% (De Munnik et al., 2015). Therefore, the genetic counselling was provided to the parents to explain the risk of recurrence in having another child.

3. Discussion

There are number of recognized genetic traits and diseases which can involve the orofacial structures. Thus, it is becoming increasingly important for a dentist to be capable of recognizing and dealing with genetic diseases. Ideally, the management of Meier-Gorlin syndrome should begin at birth. This involves

Table 2 Clinical features suggestive of Meier-Gorlin syndrome in the present case.

Primary diagnostic features	Other associated anomalies
• Overall short stature • Microtia with ear deformation • Patellar hypoplasia	• Developmental delay • Partial hearing loss • Fibrous ankylosis of TMJ • Retrognathic mandible • Reduced mouth opening • Preaxial polydactyly at right hand • Penile deviation • Ectopic kidney • Gastro-esophageal reflux • Cleft palate • Ankyloglossia • Structural brain defect

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