CASE REPORT-TREACHER-COLLINS SYNDROME

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ABSTRACT

Objectives: *To report a probable case of Treacher Collins syndrome, a rare genetic disease, to highlight the challenges in management in a resource limited setting.*

Methods: *This is the case report of GT, a 7 month old male, who presented with a history of abnormally formed ears since birth, noisy breathing and poor weight gain of 6 months duration and fever of 10days duration. He is the 5th of 5 children of a young couple of low socio economic status. On examination, he was small for age and wasted. His weight was 4.1kg which is < - 4 of the WHO Z score for child's length. He had an inspiratory stridor, intercostal and subcostal recessions. He had twisted and malformed ears, oral malocclusion, glossoptosis, macrostomia and high arched palate. He was yet to develop primary teeth. He was managed for Multiple congenital anomalies, Treacher Collins Syndrome with upper airway obstruction, and Failure to thrive. He was seen by the Ortolaryngologist and the Maxillofacial surgeon but was unable to afford the treatment and care they offered (audiological assessment, facial reconstructive surgery). Chromosomal studies could not be done due to financial constraints. He received nutritional rehabilitation, antimalarials and antibiotics. He remained in the hospital 4 days post discharge due to inability to pay the hospital fees. He has since then, not been seen at the follow up clinic.*

Conclusion: *This case is being reported to high light the importance a high index of suspicion and a knowledge of the typical dysmorphic features found in this rare condition to inform the line of patient management in a setting such as ours where, due to several confounding factors, arriving at a definitive diagnosis may be elusive.*

Key words- Treacher-Collins Syndrome

INTRODUCTION

Treacher Collins Syndrome (TCS) or Franceschetti syndrome is a rare autosomal dominant disorder, otherwise known as mandibulofacial dysostosis occuring in 1 in 50,000 persons. It presents with a characteristic facial appearance making it easily recognizable. I report a case of TCS in a 7 month old male to highlight their mode of presentation, diagnostic features, principles and challenges of management in a resource limited setting.

CASE REPORT

GT is a 7 month old male who presented with a history of abnormally formed ears since birth, noisy breathing and poor weight gain of 6 months duration and fever of 10 days duration. Mother's pregnancy period was unsupervised. However she had lower back pain in the first trimester for which she took pain relief medications. Delivery was at term at the home of a traditional birth attendant and baby cried poorly. He was predominantly breastfed for 1 month following which fortified locally prepared maize cereal was introduced. He tolerates barely 70-100 mls of pap at each feeding and is fed thrice a day. He is still being breastfed but nipple grip is poor. He is adequately immunized for age. He achieved developmental milestones at appropriate ages. He is the 5th of 5 children in a monogamous family setting. Other siblings are well. No sibling has similar features. Mother is a 25 year old petty trader with primary level of education and father a 30 year old public servant with secondary level of education.

On examination, he is small for age and wasted with thin limbs, prominent ribs and loose hanging skin over the buttock, he had an inspiratory stridor, intercostal and subcostal recessions. Weight is 4.1kg which is < - 4 of the WHO Z score for child's

length. He had twisted and malformed ears, oral malocclusion, glossoptosis, macrostomia and high arched palate. He was yet to develop primary teeth. Managing diagnosis was Multiple congenital anomalies with upper airway obstruction, Treacher-Collins Syndrome and Failure to thrive. He received nutritional rehabilitation with 3 hourly feeding via a naso gastric tube and made appreciable weight gain. He also received antimalarials and anti pyretics. Parents were however unable to do most of the requested investigations due to financial constraints. They were also unable to pay for hospital services upon discharge and so were detained in the hospital for another 4 days while soliciting funds from friends. Child was seen by the Otolaryngologist but couldn't afford an audiology assessment as requested for. He was also seen by the maxillo facial surgeons and parents told of possible facial reconstructive surgery when they have sufficient funds. After finally going home, they have not returned for follow up visits.

DISCUSSION

Treacher-Collins syndrome (TCS) is a rare autosomal dominant disease caused by a mutation of the TCOF 1 gene that has a linkage with the human chromosome 5q32 locus. Its incidence is 1:50,000 live births.[1] More than 60% of cases have no family history and so arise as de novo mutations,[1] as was the case in our patient who had no relative with similar features. Ranju et al[2] reported a case of TCS in a 10 year old girl whose father has same facial features and symptoms of TCS. Other theories suggest a possible autosomal recessive inheritance and a role for gonadal mosaicism and chromosomal rearrangement in the aetiology of the syndrome.[3]

The disease has complete penetrance, variable degree of phenotypic expression and is not associated with any form of developmental or neurologic disease.[4]The gene mutation results in an arrest in the development of facial bones. The typical features of TCS are generally bilateral symmetric anomalies of structures derived from the first and second brachial arches.[4]They include downward slanting of palpebral fissures, malar hypoplasia, mandibular hypoplasia with retrognathia, microtia, conductive hearing loss and cleft palate.[5] Other features are colobomas of the lower eyelid, paucity of lid lashes medial to the defect, congenital heart disease and cryptorchidism.[5] Abnormalities of the pinna are frequently associated with atresia of the external auditory canals with abnormalities of the middle ear ossicles, resulting in the conductive hearing loss.[6] (see table 1). Intra oral findings in our patient include a high-arched palate, malocclusion, mouth breathing, V-shaped lower arch, open bite, micrognathia and macrostomia all of which contributed to his feeding difficulties and resultant failure to thrive.

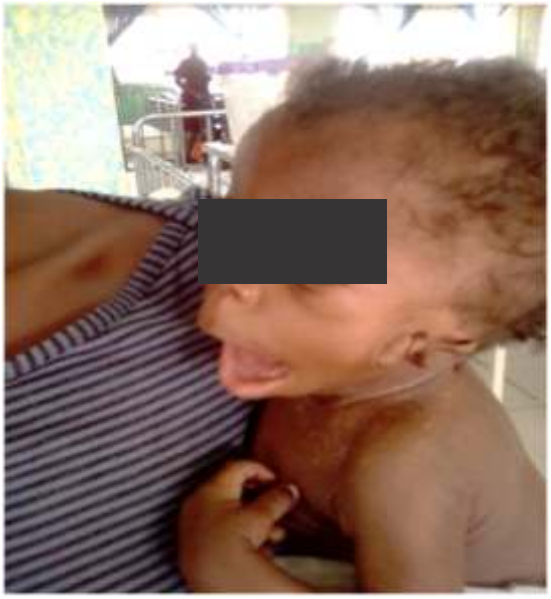
Table 1[7]

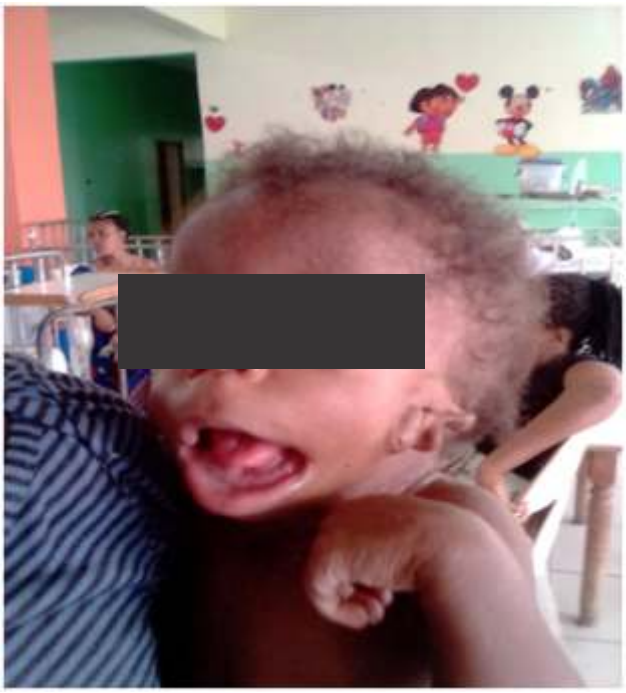
Diagnostic Clinical features of TCS	**Features absent (-) or present (+) in our patient**
Eyes- Antimongolianslant of the palpebral Fissures	-
Colobomata and hypoplasia of the Lower lids and lateral canthi	+
Hypertelorism	+
Partial absence of eyelid cilia	-
Ears- microtia	+
Conductive hearing loss	+
Hypoplasia of middle ear ossicles	-
Nose/Mouth- Nasal deformity	-
Open Bite	+
High arched palate	+
Class II or III maloclussion	+
Cleft palate + or – cleft lip	-
Facial bone formation - hypoplasia of the malar bones	+
Hypoplastic lateral aspects of orbits	+
Maxillae and mandible-	
Characteristically hypoplastic	+
Variable effects on the temperomandibular joints	+
Anterior Open Bite	+
A steep occlusal plane	+

Management of TCS is multidisciplinary involving ideally a craniofacial team, comprising of a paediatric otolaryngologist, audiologist, plastic surgeon, geneticist, psychologist, dental surgeons, and other healthcare professionals.[2,5] Their problems are usually functional and aesthetic. The cranio-facial rehabilitation of a child with TCS to tailored to the extent of the deformities involved.[5] Rao et al [8] reported a case of TCS in a 12 year old girl who had only downward slanting palpebral features and the jaw and so requiring less medical interventions than our index patient. Depending on the severity of the TCS, the patient may need some or all of the following procedures: A conductive hearing aid, correction of the cleft palate, repair of the sidewall and floor of the eye socket, repair of cheek bones, repair of eyelid notches, correction of the undeveloped jaw and chin, surgery to correct the beak-like nose, reconstruction of the ear [9] most of which are scarcely available or affordable to the average family in a resource limited setting like ours. Genetic counseling is highly recommended for affected individuals and their families as well as psychosocial support, bearing in mind the social stigma associated with such facial defect.[10]

CONCLUSION

TCS is an uncommon chromosomal disorder with a characteristic facial deformity and no neurologic deficit. Challenges in its management in a resource limited setting such as ours include availability of funds for proper radiological investigation of cranio facial defects and eventual reconstruction surgeries.





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