SYNDROMES ASSOCIATED WITH CLEFT LIP AND PALATE

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ABSTRACT

Cleft lip and cleft palate are not uncommon findings these days. Till now the Indian population considers them as a social and financial liability. Cleft lip is a common congenital anomaly that is caused by defective fusion of the medial nasal and maxillary processes during embryologic development. Cleft palate which results from failure of the lateral palatal shelves to fuse often occurs in conjunction with cleft lip, although it also may develop as an isolated defect. In the present review, we have highlighted some of the important aspects of syndromes associated with cleft lip and palate.

Key words: Cleft Lip, Cleft palate

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INTRODUCTION

In India there has been a decline in the number of deaths in children under five years of age in India from 2.5 million in 2001 to 0.98 million in 2017 but still India tops the list of countries with the largest number of under five deaths in the world. There has been a decline in the number of deaths in infant and under-5 children attributed to infectious diseases and malnutrition but the mortality attributed to congenital anomalies remains constant. Congenital oral and facial anomalies (CFA) are often a component part of congenital anomalies affecting 2-3% of all babies are a major cause of infant mortality and childhood morbidity. Syndromes or multiple anomalies are present in approximately 1% of these newborns; Syndromes are composed of multiple malformations thought to be etiologically and/or pathogenetically related. Syndromes that have cleft lip and/or cleft palate as one of the features are of interest in the quest for etiologic and pathogenetic factors, and it is estimated that 30% of cleft cases are syndromic and approximately 70% are non-syndromic.¹-³

CLEFT LIP AND PALATE

Cleft lip is a common congenital anomaly that is caused by defective fusion of the medial nasal and maxillary processes during embryologic development. Cleft palate which results from failure of the lateral palatal shelves to fuse often occurs in conjunction with cleft lip, although it also may develop as an isolated defect. Cleft Lip alone and Cleft Lip with Cleft palate are etiologically related conditions that can be grouped together as Cleft Lip with or without Cleft palate.⁴⁻⁵ Cleft palate only represents a separate entity from Cleft Lip with or without Cleft palate. In India the reported prevalence of Orofacial clefts is 1.3 per 1000 total births. The prevalence of Orofacial clefting among male births was 0.6 per 1000 total births and 0.53 per 1000 among female total births. Orofacial clefting is seen with greater frequency in a variety of specific genetic syndromes. Orofacial clefts result in a variety of problems related to appearance, feeding, speech, hearing, and socialization skills. Management involves a dedicated craniofacial team, which may include specialists in genetics, oral and maxillofacial surgery, orthodontics, otolaryngology, pediatric dentistry, pediatric medicine, plastic surgery, prosthodontics, psychology, and speech pathology.⁶

SYNDROMES ASSOCIATED CLEFT LIP AND CLEFT PALATE

Van der Woude syndrome

It is the most common form of syndromic orofacial clefting, occurring in 1 out of every 40,000 to 100,000 births. It is estimated that 2% of all Cleft lip and Cleft Palate cases are part of van der Woude syndrome, which is caused by mutations of the gene that encodes interferon regulatory factor 6 (IRF6). Associated symptoms are paramedian lip pits, hypodontia and abnormal salivary gland morphology.⁷
2. Popliteal pterygium syndrome

It is the allelic variants of Van der Woude syndrome that is they both are caused by different mutations of the same gene. Popliteal pterygium syndrome includes Van der Woude syndrome features plus popliteal pterygium, syngnathia, distinct toe or nail abnormality, syndactyly and genito-urinary malformations. It is the allelic variants of Van der Woude syndrome that is they both are caused by different mutations of the same gene.

2. Median facial dysplasia

It is associated with deficient mid facial structures along with cleft lip and cleft palate. Deficient mid facial structures leads to dish face maxillary hypoplasia and Class III malocclusion.

3. Pierre Robin syndrome

It is the sequence type of abnormality in which there is micrognathia, glossoptosis and airway obstruction are cardinal features along with cleft palate. Genes involved in this syndrome are 2,4,11,17.

4. Kabuki syndrome:

It was first identified and described in 1981 by two Japanese groups, led by scientists Norio Niikawa and Yoshikazu Kuroki occurring in 1out of 32900 births demonstrates an X-linked dominant pattern of inheritance. Clinical manifestations other than cleft palate are postnatal growth deficiency, intellectual disability, skeletal abnormalities involving spine, facial features like elongated palpebral fissures, arched and broad eyebrows, short columella and large, prominent, ears.

5. Velocardiofacial syndrome

It is associated with deletion of chromosomal material from long arm of chromosome 22 characterized by congenital cardiac defects, cleft palate, velopharyngeal insufficiency, distinct facial features, immunological problems, learning disabilities, and psychological disorders.

6. Treacher Collins syndrome

The syndrome is named after Edward Treacher Collins, an English surgeon and ophthalmologist, who described its essential traits in 1900. This syndrome is autosomal dominant characterized by breathing problems, vision problems, cleft palate, and hearing loss.

Management:

In rehabilitation of patient of cleft palate various prosthesis have been used

1. To facilitate feeding:
   - Prosthesis has been designed to prevent nasal regurgitation, entrance of tongue into the defect.
   - It also prevents otitis media and other pharyngeal infections.
   - Obturators can be used as feeding obturators or they can be provided after surgery to prevent any communication between oral and nasal cavity.

2. To facilitate speech:
• Prosthesis has been provided when soft palate is insufficient leading to escape of air to nasal cavity.
• Prosthesis consist of two parts palatal part and pharyngeal part to extend posteriorly to close velopharyngeal space thus aid in speech.

3. To replace missing teeth:
• Fixed partial, removable partial even implants could be used to replace the missing teeth.10

CONCLUSION
The facial and dentofacial anomalies have been a subject of major concern in India till date. Though the number has gone down but India still has a lot to work upon. Till now the Indian population considers them as a social and financial liability. The numbers can further be decreased with the help of proper pre natal genetic medical investigations and genetic counseling.

REFERENCES
1. WHO report on congenital malformations 2017
2. Shammas M, Arya PS, Viji A Thottumkal, MG Deepak: Congenital anomalies a major public health issue in India; IJPC 2013, 3(3), 577-585.