Role of MSX1 Gene in Orofacial Clefting: A Systematic Review

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INTRODUCTION

Orofacial clefting has always been the bane of the human race. It affects the overall wellbeing of the affected individual both physically as well as physically. The incidence of orofacial clefts has been around 1:500. Orofacial clefts can occur from an isolated cleft palate to a bilateral cleft lip and palate. Severe facial deformation gives the affected individuals a psychological set back. In developing countries like India where the cleft care does not reach to the rural population, the severity of unoperated clefting becomes more evident.

The search for cause of orofacial clefting has been under research for a while now. While two forms of orofacial clefting, syndromic and non-syndromic clefting have been researched extensively. The interest in non-syndromic clefting has grown more as it is a condition where an apparently healthy individual shows orofacial clefting without any other systemic condition.

In 1969 Carter proposed a model (MF/T) multifactorial clefting inheritance, where he stated that non-syndromic clefting was caused by the additive effects of minor abnormal genes and environmental factors.

MSX genes

The MSX group of genes in vertebrates comprise of a small family of chromosomally unlinked homeobox genes related to the Drosophila muscle segment homeobox (MSH). MSX genes are expressed in vertebrate specific tissues, including sensory placodes, neural crest, bone and teeth. The MSX genes are classified into MSX1 and MSX2. MSX3 found in mice is placed as a subclass of MSX1 subclass.

Knockout experiments with mice have shown a link to MSX1 to failure to form teeth, and craniofacial abnormalities including absence alveolar bone in the jaws and disturbances in the formation of the parietal, nasal, frontal, cleft palate and malleus of middle ear.

Objective

To test the null hypothesis
MSX1 gene mutation causes orofacial clefting.

Methodology

Three search bases, Pubmed, Science direct and Cochrane were searched using the key words. The Inclusion criteria for the study was Direct association of MSX1 gene mutation to orofacial clefting. Human subjects with Cleft
RESULTS

Pubmed showed 5, Science direct 79 and Cochrane 1 articles.

Further using the inclusion criteria, 5 articles were selected.

DISCUSSION

The Role of MSX 1 gene in Human orofacial clefting has always been debated, this systematic review was designed to find if there were any human studies that implicated orofacial clefting to the MSX 1 gene. In all 4 studies have implicated a direct relation to orofacial clefting. These studies have covered most of the populations around the world. AC Lidral and BC Reising found a Met61Lys substitution in two siblings in a big family with autosomal-dominant tooth agenesis. Venkatesh S Prasad and Venkatesh Shivani found a novel mutation (414G to T) in a south Indian population. Seishi Yamaguchi et al found two MSX1 variants with an amino acid substitution; Thr174Ile (T174I) of a hypodontia case and Leu205Arg of a familial oligodontia case in a Japanese Population. Derya Ceyhan, Zuhal Kirzioglu and Nilufer Sahin Calapoglu reported mutations in the MSX 1 gene from a predominant Turkish population.

CONCLUSION

MSX1 gene which is a homeobox gene has been implicated in the formation of orofacial clefting. Several human studies have shown mutations in different populations.

REFERENCES