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Genetic Medicine In A Multi-disciplinary Cleft Clinic At A Tertiary Care Center: A 7 Year Experience

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We describe our 7 year experience in the multi-disciplinary Cleft Clinic at The Johns Hopkins Hospital. As has been previously demonstrated and increasingly recognized, patients and families affected by orofacial clefting and/or other craniofacial anomalies can benefit from comprehensive and coordinated evaluation and management by multiple subspecialties. At our institution, this clinic combines the expertise of pediatric plastic surgery, speech and language pathology, pediatric dentistry and orthodontia, pediatric otolaryngology, audiology, and genetics.With institutional review board approval, we are performing a 7 year retrospective analysis of all patients assessed by genetic medicine professionals in our multidisciplinary cleft clinic. Since 2012, there have been over 1,200 encounters for an initial or continuing genetic evaluation of more than 670 families. The primary objective of this analysis is to determine if there is overrepresentation in our cleft clinic as compared to the general population of the following patient characteristics: conception via assisted reproductive technologies; in utero exposure to one or more teratogenic agents; and/or adoption. As a secondary objective, we will also determine if orofacial clefting, in general and by type (i.e., palate alone, lip and palate, lip alone), is associated with advanced parental age, the presence of other congenital anomalies (e.g., congenital heart defect, craniosynostosis), poor growth, and/or a positive family history of orofacial clefting. Finally, we will define the yield of our genetic medicine evaluation to differentiate syndromic from non-syndromic orofacial clefting in this patient population. Through directed cytomolecular testing, we are often able to establish a specific genetic diagnosis for many of these patients. Several of these cases will be presented, representing different testing modalities including SNP microarray, single and multi-gene sequencing and deletion/duplication analysis, as well as clinical exome sequencing.Reaching a diagnosis has tremendous value for a patient and their family as we are able to improve anticipatory guidance and refine recurrence risk estimates. Examination of this large and unique dataset will also contribute to our understanding of orofacial clefting and demonstrate the value of multi-disciplinary care for these complex patients.

Comments (un-moderated)

I would like to echo this sentence: "Reaching a diagnosis has tremendous value for a patient and their family as we are able to improve anticipatory guidance and refine recurrence risk estimates."
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