Van der Woude syndrome

A four-month-old healthy male child, first born of a non-consanguineous marriage, presented with lower lip pits (*Fig. 1*). On examination, there was no cleft lip, cleft palate, submucous cleft palate or cleft uvula. There were no popliteal webbing, syndactyly, syngnathia or genital and nail anomalies. His milestones were normal. There was no family history of similar condition. A diagnosis of Van der Woude syndrome (VWS) was made. The child was to undergo surgical removal of the pits (fistulas) at the age of two years.

VWS is a rare autosomal dominant cranio-facial disorder characterized by paramedian lower lip pits (fistula labii inferioris congenita), cleft palate and or cleft lip. VWS is a form of syndromic orofacial clefting with very high penetrance and varied expressivity. The clinical features described may occur in any consisation. Children with VWS have normal intelligence. Congenital lip pits are characteristically present in 80% of cases and are usually connected with heterotopic salivary glands; occasionally, salivation may occur. Other less frequent features include cleft uvula and hypodontia (missing central and lateral incisors, canines, and/or bicuspids). The interferon regulatory factor 6 gene responsible for this disorder has been mapped to the long arm of chromosome 1 at q32-41. Since lesser expressions of Van der Woude syndrome are common it is important to recognize this condition early because of the genetic implications in counseling families about cleft lip or cleft lip and/or palate.

Popliteal pterygium syndrome (PPS) is another rare autosomal dominant disorder with a similar orofacial phenotype but it includes skin and genital anomalies. The main clinical manifestations of PPS are popliteal webbing (90%), cleft palate with or without cleft lip (90%), and genital and nail anomalies (51%), lower lip pits (46%), syngnathia (43%), and syndactyly. Phenotypic overlap and linkage data suggest that these two disorders are allelic.

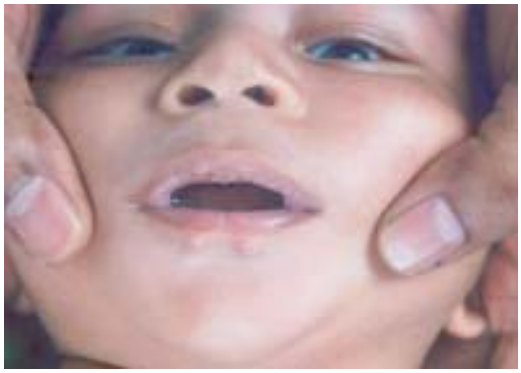

Fig. 1. Bilateral symmetrical lip pits present close to the vermilion border of the lower lip about 0.5 cm off the midline.

Acknowledgement

The authors thank Dean, Dr. M.E. Yeolekar, for granting us permission to publish this clinical image.

Contributors: SK was responsible for diagnosis of the case, drafting the article and will act as the guarantor for the article; SP for review of data and discussing core ideas.

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