FAMILY STUDY OF AURICULO-CONDYLAR SYNDROME.

OBJECTIVE: Auriculo-condylar syndrome (ACS, OMIM 602483) is a rare craniofacial deformity syndrome caused by improper development of the first and second branchial arches. It is transmitted in an autosomal dominant manner. Deformities involve prominence of the ears, marked constriction between the lower and middle thirds of the pinna and abnormalities of the mandible including micrognathia. There is typically no hearing loss or abnormalities of the ossicles. However associations have been noted with an abnormal palate, hypotonia, and neurodevelopmental delay. In total, this defect results in impairment of breathing, eating, speech, and cosmesis. To date, only small families with parent-child transmission have been reported. Here we present a large family with four generations of affected individuals. Variable expressivity and penetrance are exhibited. Power analysis demonstrates that we have greater than a 95% chance of identifying a locus with a genome wide screen. A genome wide scan using Weber linkage markers has been initiated to identify potential loci involved in the pathogenesis of this syndrome.