

Evidence of a major gene in cleft lip/palate susceptibility by means of segregational analysis in the Chilean population Evidencia de un gen mayor en la susceptibilidad a la fisura labiopalatina mediante analisis segregacional en la población Chilena.

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The most generally accepted model for cleft lip/palate not associated to specific syndromes has been the one that postulates multifactorial inheritance with a threshold. Recent studies using complex analytical techniques have suggested the existence of a major gene with decreased penetrance in its etiology. Some authors have postulated that only a fraction of all cases of non-syndromic CL (P) would be explained by a single major gene. Other cases may be due to different genes, to environmental agents or to the interaction between them. The present study tests the monogenic inheritance model for CL (P) using segregation analysis in a sample of 211 extended pedigrees collected through CL (P) affected probands. The hypotheses of an autosomic recessive gene was analyzed in 151 sibships (size 2 or more) using "Apert's" method, "maximum likelihood method", "singles method" and "sib's, method" (single incomplete ascertainment). Results obtained do not support the hypotheses of autosomal reces