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CLINICAL PREDICTORS OF VELOCARDIOFACIAL SYNDROME AS REPORTED IN PATIENTS' CHARTS

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OBJECTIVE: The velocardiofacial syndrome (VCFS) involves a deletion of part of chromosome 22 and is associated to more than 180 clinical conditions. With a high variation in clinical findings, early identification of this condition may be difficulty. The objective of this study was to compare clinical findings reported in medical charts in two groups of patients with VCFS: with positive molecular testing and without testing but with clinical sigs of VPI. METHODS: A list of patients with VCFS was identified in a large craniofacial center. The first 40 patients with signs of VPI that could be grouped according to presence or absence of molecular testing were included in the study. RESULTS: The charts from 20 patients without molecular tests, 20 patients with positive molecular test for the deletion and 2 with negative molecular test were reviewed. A total of 17 health professional areas reported signs of VCFS in one or more charts, with speech pathologists reporting in all charts, clinical geneticists in 98% and pediatrician in 83% of the charts studied. A list of 31 clinical signs were reported by these professionals. Cardiac and respiratory conditions and behavioral problems were more prevalent in the group with positive molecular test. Incomplete cleft palate was reported for 43% of the patients, submucous cleft palate for 26%, congenital velopharyngeal dysfunction for 19% and other anomalies for 12%. CONCLUSION: Patients with VCFS that are treated in a Cleft Palate Center are biased to presenting with cleft palate or velopharyngeal dysfunction. Presence of cardiac and respiratory conditions and behavioral problems in this population can be a sign of VCFS.