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Congenital Heart Defects and Dysmorphic Facial Features in Patients Suspicious of 22q11.2 Deletion Syndrome in Southern Brazil

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Abstract

22q11.2 deletion syndrome (22q11.2DS) is considered one of the most frequently observed chromosomal abnormalities in association with congenital heart disease (CHD), which can also include some combination of other features. Thus, the aim of this work was to verify the profile of dysmorphic features and heart defects found in patients referred to a reference center in Southern Brazil with clinical findings suggestive of 22q11.2DS. In the overall sample group, only patients with dysmorphic facial features (skull, eyes, ear, and nose) associated with CHD (obstructive pulmonary valve ring, truncus arteriosus, and bicuspid aortic valve associated with atrial septal defect and/or right aortic arch) had a 22q11.2 deletion. These findings proved to be reliable clinical criteria for referral to perform fluorescent *in situ* hybridization investigation for 22q11.2 deletion.

Keywords: 22q11 deletion syndrome; congenital; facial dysmorphism; heart defects.

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