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THE CLINICAL AND NEUROCOGNITIVE FEATURES OF DIGEORGE SYNDROME PATIENTS OF CENTRO DE GENÉTICA MÉDICA DOUTOR JACINTO MAGALHÃES

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Introduction: The 22q11 deletion occurs in the presence of monosomic deletions of the long arm of chromosome 22, which can vary and alter the phenotype of the disease, and affects 1 in 2,000 – 4,000 live births. DiGeorge's syndrome (DGS)/velocardiofacial syndrome is one of these possible phenotypes under 22q11DS umbrella term. In order to classify the phenotypical features of DiGeorge's syndrome (DGS), also referred as velocardiofacial or 22q11.2, it was created the CATCH22, an acronym that sums all the major clinical features of the deletion (Cardiac abnormality, Abnormal facies, Thymic aplasia, Cleft palate and Hypocalcemia/Hypoparathyroidism). Additionally, features like immunodeficiency, feed-swallow abnormality, skeletal problems and developmental delay are also described as major clinical symptoms of this deletion. Regarding cardiac malfunctioning, the most common abnormalities are tetralogy of Fallot, interrupted aortic arch, ventricular septal defect and truncus arteriosus.

Methods: Twenty four clinical cases of DGS were consulted in order to create a data base of all clinical and neuropsychological information.

Results: In our sample of 24 subjects diagnosed with DGS with ages between 12 and 52 years, we can in fact consider the congenital heart defect as the major symptom (82%, 20/24), with ventricular septal defect and tetralogy of Fallot as the most prevalent ones. Although they are absent from the most common heart defects, 18% (4/24) of our sample have other cardiac symptoms. In our sample, 13 subjects were evaluated cognitively reporting highly variable values, since moderate (38%, 5/13) with IQ's between 51 and 69; 31% (4/13) has a borderline IQ value, between 71 and 79; and 31% (4/13) of patients have normal IQ values, which is demonstrative of the high variability of the neurocognitive profile of this sample. In adults and adolescents, we can also register the development of psychiatric profiles related with schizophrenia, hyperactivity and other schizoaffective and behavioural disorder.

Conclusions: Clinically the pattern of cardiac abnormality prevalence is preponderant over all the other typical features of the disease and therefore these cardiac features are clinically subject to follow-up studies. Beside these physiological malfunctions, this deletion has also associated a neurocognitive phenotype, characterized by developmental delay in early stages and evolution to psychiatric dysfunction. However,

the clinical registers show that there is a great need but poor clinical following of these patients by a multidisciplinary team, especially in the cognitive and behavioural deterioration that can occur during the lifespan, which would prevent psychiatric manifestations and improve quality of life.