# PE 128 - CLINICAL AND ANTHROPOMETRIC CHARACTERIZATION OF 100 BRAZILIAN PATIENTS WITH 22Q11.2 DELETION SYNDROME

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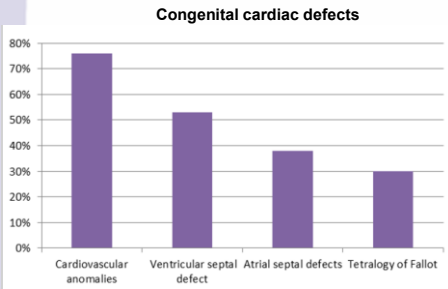
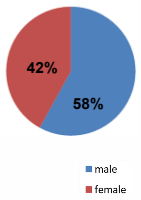
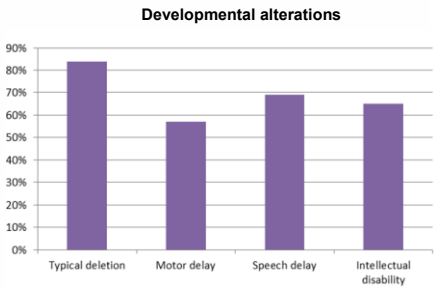
**INTRODUCTION:** The 22q11.2 deletion syndrome is a multisystem genetic disorder resulting from a hemizygous microdeletion, with a prevalence of approximately 1:4,000 live births. This study provides a comprehensive clinical and anthropometric profile of Brazilian patients, offering insights into the phenotypic diversity and health challenges associated with this condition.

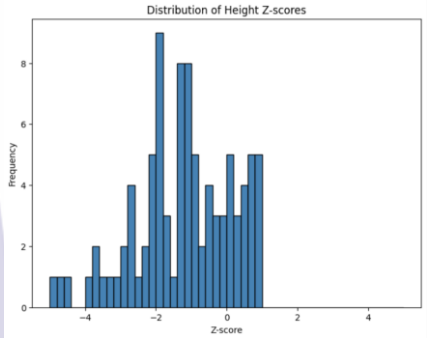
**METHODS:** We conducted a retrospective review of medical records spanning 2000–2024 from a single specialized genetic center in Brazil. Clinical data, confirmed primarily via MLPA and occasionally by FISH, were analyzed according to CDC growth standards. Anthropometric parameters and clinical findings were systematically evaluated. None of the authors have any conflict of interest for this paper.

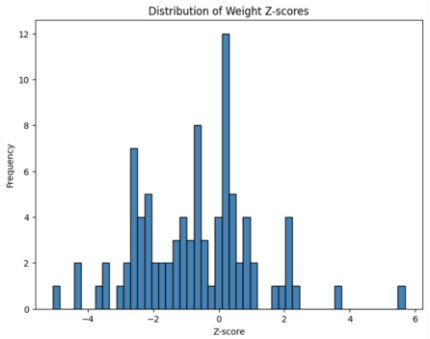
**RESULTS:** Among the 100 patients analyzed (58% male, 42% female), the average age at clinical diagnosis was 6.46 years (0-40 years). The majority (84%) presented with the common 3 Mb deletion and 16% had a atypical deletion size. Cardiovascular anomalies were the predominant feature (76%), with ventricular septal defects (53%) and tetralogy of Fallot (30%) being most prevalent. Developmental challenges were common, including speech delay (69%), motor delay (57%), and intellectual disability (65%). 17% of cases lacked congenital anomalies at birth, with symptoms manifesting later in life. Other clinical presentations were analysed: 36% had a nasal voice, with nine patients with cleft lip and palate; 27% had neonatal hypocalcemia, and 29% experienced hypocalcemic episodes after their first year of life. Key findings included short stature (26%, mean height z-score: -1.2), weight variability (28% underweight, 7% overweight), and microcephaly (19%). 74% had normal stature for their age.




**CONCLUSIONS:** This cohort represents the largest single-center dataset on 22q11.2 deletion syndrome in Brazil, highlighting phenotypic consistencies and unique regional patterns. Although the focus was on characterization of anthropometric data, frequencies of other typical conditions of the syndrome are also described, with prevalences similar to other studies. These findings contribute to the global understanding of this syndrome, emphasizing the need for early diagnosis and tailored management strategies.

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