

Prevalence of clinical features of Aarskog-Scott Syndrome

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Abstract

Aarskog-Scott syndrome is a genetically and rare condition originate by a mutation in FGD1 gene. In current literature a several numbers of clinical features classification were dispoñible. A standardization of this feature promotes a better orientation to clinical participants in diverse fields such as medicine and dentistry. To assess the prevalence of this clinical signs described in literature, a systematic review was carried out. 182 studies were screened and after a criterial appraisal, 22 studies were considered for qualitative analysis. The main inclusion criteria are studies that show a genetical test results of presented cases and complete phenotypical description. The results shows that craniofacial and orthopaedic corresponds to the highest scores of prevalence. 52 different pathogenic variants were described among the 58 cases, in which 33 patients presented with a point pathogenic variant (missense or nonsense mutations), representing about 56% of the total. In addition, 14 small deletions, four small insertions, three gross deletions, three splicing, and one duplication were described. Craniofacial manifestations were the most frequently described in the literature, representing 38.8%, orthopaedic changes represented 18.1%, ophthalmological and neurological manifestations together comprised 25.8%, genitourinary system corresponded to 8.6% The aim of this study is discussing and show the novel clinical features classification and promotes an evidence based clinical genetics pratice.

Biography

Academic of the third year of Medicine at the Metropolitan Institute of Higher Education / University of Ipatinga, MG. He does scientific initiation at the Federal University of Minas Gerais (UFMG) in the Pediatrics Department of FMUFG in the Research Group on Medical Education headed by the PhD. Monitor in the Program of Neuroanatomy and Anatomy of the Osteomuscular, Endocrine, Urinary, Male and Female Genital Systems beginning in 2019. He was Vice-President of the Academic League of Neurology and Neurosciences and Scientific Director of the Academic League of Intensive Medicine. Approved and completed the Yale School of Medicine course in anatomy of the chest, abdomen and pelvis taught by Professor Charles Duncan in 2020. Participate in the IMES Child Life extension project specializing in basic life support for lay people and pediatric emergencies.