22q11.2 Distal Deletion Syndrome

22q11.2 distal deletion syndrome and DiGeorge syndrome are both genetic disorders caused by deletions on chromosome 22q11.2, but they differ in the specific regions affected and their clinical presentations. DiGeorge syndrome (DGS), also known as 22q11.2 deletion syndrome, typically involves a deletion between low-copy repeats (LCRs) A and D on chromosome 22q11.2. This deletion results in a constellation of clinical features including congenital heart defects, thymic hypoplasia leading to immunodeficiency, hypoparathyroidism with resultant hypocalcemia, and characteristic facial features. The syndrome is highly variable, with additional manifestations such as developmental delays, psychiatric disorders, and renal anomalies.[1-4]

22q11.2 distal deletion syndrome, on the other hand, involves deletions that are located distal to the common 3 Mb deletion region associated with DGS. These deletions are typically flanked by LCR22-4 and either LCR22-5 or LCR22-6. Patients with distal deletions present with a different set of clinical features, which may include facial dysmorphisms, growth delays, developmental delays, and mild skeletal abnormalities. Cardiovascular malformations and cleft palate can also occur, but the overall phenotype is distinct from that of DGS.[5]

In summary, while both syndromes involve deletions on chromosome 22q11.2, DiGeorge syndrome is characterized by deletions between LCR22-A and LCR22-D and presents with a broader spectrum of systemic involvement, whereas 22q11.2 distal deletion syndrome involves deletions distal to the common DGS region and presents with a distinct clinical phenotype.[5]

Resources

1. Medline Plus
2. Gene Reviews
3. 22q11.2 Deletion Syndrome. McDonald-McGinn DM et al. Nat Rev Dis Primers 2016;1:15071
4. Chromosome 22q11.2 deletion syndrome and DiGeorge syndrome. Sullivan KE. Immunol Rev. 2019;287(1):186-201
5. 22q11.2 distal deletion: A Recurrent Genomic Disorder Distinct from DiGeorge Syndrome and Velocardiofacial Syndrome. Ben-Shachar, S et al. American Journal of Human Genetics. 2008;82:214-221

syndrome