Disorders related to Acrodysostosis

Symptoms of the following disorders can be similar to those of acrodysostosis. Comparisons may be useful for a differential diagnosis.

Albright hereditary osteodystrophy (AHO)

Albright hereditary osteodystrophy (AHO) is a rare disorder characterized by short stature, an unusually round face, abnormally short fingers (brachydactyly), and/or the development of bony growths (osseous plaques) on the surface of the skin but not in the deep connective tissue. These growths may spread to the lower level of the skin as well (subcutaneous ossification).

Other symptoms may include mild intellectual disability and obesity.

AHO may be isolated or associated with hormone resistance, such as parathyroid hormone resistance which manifests as abnormally low levels of calcium in the blood (hypocalcemia).

Therefore, symptoms of pseudohypoparathyroidism include weakness, muscle cramps, excessive nervousness, headaches, and/or abnormal sensations such as tingling, burning, and numbness of the hands. The association of AHO and hormone resistance is termed pseudohypoparathyroidism type 1A.

AHO (sometimes called pseudopseudohypoparathyroidism or PPHP) and PHP1A are caused by loss of function mutations of the same gene (GNAS). GNAS encodes the alpha stimulatory subunit of the G-proteins that are needed to properly respond to parathyroid hormone and other hormones.

Each condition can be inherited in an autosomal dominant manner. However, isolated AHO (PPHP) is inherited from fathers whereas PHP1A is inherited from mothers.

5q12.1 Haploinsufficiency Syndrome

5q12.1 Haploinsufficiency Syndrome is an extremely rare disorder that has only been described in several individuals.

These individuals have structural chromosome abnormalities (e.g. deletions) that involve the PDE4D gene, resulting in half the normal production of the protein product of that gene (haploinsufficiency). The symptoms of these individuals were extremely similar to those seen in individuals with **acrodysostosis type 2** including underdevelopment of certain facial bones, brachydactyly, and intellectual disability.

2q37 Microdeletion Syndrome

2q37 Microdeletion Syndrome is a rare disorder characterized by a broad range of signs and symptoms.

Affected individuals often develop varying degrees of intellectual disability, abnormal short bones in the fingers and hands (brachymetaphalangy), short stature, obesity, and distinctive facial features.

Additional symptoms include diminished muscle tone (hypotonia), joint abnormalities, abnormal sideways curvature of the spine (scoliosis), and autism spectrum disorder.

Some affected individuals may have congenital heart disease, seizures, central nervous system abnormalities, hernias, gastrointestinal abnormalities, and kidney (renal) malformations. Parathyroid hormone resistance was described in few cases.

2q37 microdeletion syndrome is caused by a small loss of genetic material on the long arm (q) of chromosome 2. The specific gene(s) involved in this disorder are not known.