

Conradi-Hünemann syndrome is a rare genetic disorder characterized by skeletal malformations, skin abnormalities, cataracts and short stature. The specific symptoms and severity of the disorder may vary greatly from one individual to another. Conradi-Hünemann syndrome is classified as a form of chondrodysplasia punctata, a group of disorders characterized by the formation of small, hardened spots of calcium on the "growing portion" or heads of the long bones (stippled epiphyses) or inside other areas of cartilage in the body. Conradi-Hünemann syndrome is commonly associated with disproportionate and asymmetric shortening of long bones, particularly those of the upper arms (humeri) and the thigh bones (femora), curvature of the spine and mild to moderate growth deficiency, resulting in short stature. Many affected individuals also have a prominent forehead; unusually flattened midfacial regions (midfacial hypoplasia), with a low nasal bridge; loss of transparency of the lenses of the eyes (cataracts); sparse, coarse scalp hair; and/or abnormal thickening, dryness, and scaling of the skin. Conradi-Hünemann syndrome is inherited as an X-linked dominant trait that occurs almost exclusively in females.