

Hypochondroplasia is a genetic disorder characterized by small stature and disproportionately short arms, legs, hands, and feet (short-limbed dwarfism). Short stature often is not recognized until early to mid childhood or, in some cases, as late as adulthood. In those with the disorder, bowing of the legs typically develops during early childhood but often improves spontaneously with age. Some affected individuals may also have an abnormally large head (macrocephaly), a relatively prominent forehead, and/or other physical abnormalities associated with the disorder. In addition, in about 10 percent of cases, mild mental retardation may be present. Hypochondroplasia appears to affect females and males in relatively equal numbers. The features of the disorder were originally reported in 1913; hypochondroplasia was described as a distinct disease entity in 1924. Over 100 cases have since been recorded in the medical literature, including isolated (sporadic) and familial cases. Hypochondroplasia is thought to have an incidence of approximately one-twelfth that of achondroplasia. (Incidence refers to the number of new cases of a particular disorder or condition during a specific period.) The estimated frequency of achondroplasia has ranged from about one in 15,000 to one in 35,000 births. (For further information on achondroplasia, please see the "Related Disorders" section of this report below.) As noted previously, in individuals with hypochondroplasia, short stature often may not be recognized until early or mid childhood or as late as adulthood. The disorder may be diagnosed based upon thorough clinical examination; identification of characteristic physical findings (e.g., short stature, brachydactyly, genu varum, macrocephaly); x-ray studies; and/or other diagnostic techniques.