

Kenny-Caffey syndrome type 2 (KCS2) is an extremely rare hereditary skeletal disorder characterized by thickening of the long bones, thin marrow cavities in the bones (medullary stenosis), and abnormalities affecting the head and eyes. Most cases are obvious at birth (congenital). The primary outcome of KCS2 is short stature. Intelligence is usually normal. Individuals with KCS may also have recurrent episodes of low levels of calcium in the blood stream (hypocalcemia) that is caused by insufficient production of parathyroid hormones (hypoparathyroidism). In most cases, KCS2 is an autosomal dominant genetic disorder. KCS2 is an extremely rare skeletal disorder that affects males and females in equal numbers. Fewer than 60 cases have been reported in the medical literature. Onset of hypocalcemia is usually within two to three months of life; the hypocalcemia is not permanent (transient). In an adult, episodes of hypocalcemia may be due to stress or follow surgery or illness. KCS2 was first described in the medical literature in 1966. The diagnosis of KCS2 may be confirmed by x-ray studies of the skeleton that reveal distinctive thickening of the outer layers (cortexes) of long bones along with unusually thin marrow cavities. Blood tests can detect episodes of low levels of calcium in the blood (hypocalcemia).