Jansen type metaphyseal chondrodysplasia is an extremely rare disorder that affects males and females in equal numbers. Approximately 20 cases have been reported in the medical literature. The diagnosis of Jansen type metaphyseal chondrodysplasia may be suspected during infancy or early childhood. The diagnosis may be confirmed by a thorough clinical evaluation, identification of characteristic physical findings, and a variety of specialized tests, particularly advanced imaging techniques. These techniques include x-ray studies that may reveal abnormal development of the large (bulbous) ends (metaphyses) of certain bones of the body, particularly those of the arms and legs. Laboratory tests that detect abnormally high levels of calcium in the urine (hypercalciuria) and blood (hypercalcemia) are helpful in confirming the diagnosis.