Hereditary hyperphosphatasia is a rare genetic bone disorder (osteopathy) that usually becomes apparent during infancy or early childhood. Affected individuals develop progressive skeletal malformations especially in the long bones of the arms and legs. Skeletal malformations in the legs may cause problems walking and may eventually result in short stature. Additional symptoms include pain, fractures of affected bones, and muscle weakness. Because the biochemical and radiographic findings of hereditary hyperphosphatasia are similar to those of Paget's disease (a focal skeletal disorder of adults characterized by abnormal bone turnover), the disorder is sometimes referred to as juvenile Paget's disease. However, despite these similarities, the two disorders are distinct. Hereditary hyperphosphatasia is inherited in an autosomal recessive pattern. Hereditary hyperphosphatasia affects males and females in equal numbers. Like all recessive disorders it is more common in countries where within-family marriage is practiced. More than 50 cases have been described since the disorder was first reported in the medical literature in 1956. A diagnosis of hereditary hyperphosphatasia is made based upon a thorough clinical evaluation, identification of characteristic symptoms and a variety of x-rays tests that reveal distinct radiographic findings. Affected individuals also have elevated levels of serum alkaline phosphatase and other biochemical markers of bone turnover, detectable through blood and urine tests.