Campomelic syndrome is a rare congenital disorder in which multiple anomalies are present. It is characterized by bowing and angular shape of the long bones of the legs, especially the tibia; multiple minor anomalies of the face; cleft palate; other skeletal anomalies such as abnormalities of the shoulder and pelvic area and eleven pairs of ribs instead of the usual twelve; underdevelopment of the trachea; developmental delay in some cases and incomplete development of genitalia in males such that they appear to be females. Campomelic syndrome is a rare disorder that is thought to affect females twice as often as males. These numbers may not be accurate as some patients with this disorder have associated sex reversal and have been mistakenly identified as the opposite sex. Approximately one hundred cases of this disorder have been reported in the medical literature. Diagnosis is based on clinical examination, x-rays of vertebrae, hips, chest, legs and feet, ultrasound of kidneys and echocardiogram of the heart. DNA analysis of blood can confirm a mutation in the SOX9 gene.