"Leri Pleonosteosis is an extremely rare inherited disorder characterized by unusual, flattened facial features, abnormalities of the hands and feet, skeletal malformations, short stature, and/or limitation of joint movements. Characteristic abnormalities of the hands and feet may include unusually broad and/or short thumbs and great toes (brachydactyly) that may be bent outward from the body (valgus position); as a result, the hands may have a ""spade-shaped"" appearance. Skeletal malformations may include knees that are bent backward (genu recurvitum) and abnormal enlargement of the cartilaginous structures that surround the upper portion of the spinal cord (posterior neural arches of the cervical vertebrae). In addition, affected individuals may develop thickened tissue on the palms (palmar) and forearms. Symptoms may vary from case to case. Leri pleonosteosis is inherited as an autosomal dominant genetic trait. Leri pleonosteosis is an extremely rare inherited disorder that affects males and females in equal numbers. Approximately 20 cases have been reported worldwide, with most of these outside of North America. The symptoms and physical characteristics associated with Leri pleonosteosis usually become apparent during infancy or early childhood. The diagnosis of Leri pleonosteosis may be established by a thorough clinical evaluation, characteristic physical findings, detailed patient history, and/or specialized tests including advanced imaging techniques (e.g., various x-ray methods). For example, enlargement of the cartilage that surrounds the upper spinal cord (posterior neural arches of the cervical vertebrae), is an important characteristic of this disorder and is potentially detectable by x-ray studies."