Marfanoid habitus-inguinal hernia-advanced bone age syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Marfanoid habitus-inguinal hernia-advanced bone age syndrome. ORPHA:314041

Marfanoid habitus-inguinal hernia-advanced bone age syndrome is a very rare developmental defect with connective tissue involvement disorder characterized by tall stature, inguinal hernia, facial dysmorphism (including a long, triangular face, prominent forehead, telecanthus, downslanting palpebral fissures, bilateral ptosis, everted lower eyelids, large ears, long nose, full, everted vermilions, narrow and high arched palate, dental crowding), and radiologic evidence of advanced bone age. Additional manifestations include hyperextensible joints, long digits, mild muscle weakness, myopia, and foot deformities (i.e. hallux valgus, talipes equinovarus).