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SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. [SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome](#). ORPHA:157965

Ehlers-Danlos syndrome, spondylocheiroadysplastic type is a subtype of Ehlers-Danlos syndrome characterized by skeletal dysplasia comprising platyspondyly with moderate short stature, osteopenia and widened metaphyses, in addition to hyperextensible, thin, easily bruised skin, hypermobility of small joints with tendency to contractures, prominent eyes with bluish sclerae, wrinkled palms, atrophy of the thenar muscle and tapering fingers.