

Open Peer Review on Qeios

Chondrodysplasia with joint dislocations, gPAPP type

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Chondrodysplasia with joint dislocations, gPAPP type</u>. ORPHA:280586

Chondrodysplasia with joint dislocations, gPAPP type is a rare, genetic, primary bone dysplasia characterized by prenatal onset of disproportionate short stature, shortening of the limbs, congenital joint dislocations, micrognathia, posterior cleft palate, brachydactyly, short metacarpals and irregular size of the metacarpal epiphyses, supernumerary carpal ossification centers and dysmorphic facial features. In addition, hearing impairment and mild psychomotor delay have also been reported.

Qeios ID: AVR7AZ · https://doi.org/10.32388/AVR7AZ