DISEASE:
Chondrodysplasia with joint dislocations, gPAPP type

NAME:	Chondrodysplasia with joint dislocations, gPAPP type
DESCRIPTION:	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.
ORPHACODE:	280586
SYNONYMS:	gPAPP deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	BPNT2
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RELATED CONTENT

Related Genetic Tests

- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL

Related Analytes

- 3'(2'), 5'-bisphosphate nucleotidase 2

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL