DISEASE:
Multiple epiphyseal dysplasia, Al-Gazali type

NAME:	Multiple epiphyseal dysplasia, Al-Gazali type
DESCRIPTION:	A rare primary bone dysplasia characterized by the association of multiple epiphyseal dysplasia with macrocephaly and dysmorphic facial features (such as frontal bossing, hypertelorism, flat malar region, low-set ears, and short neck). Patients are of normal stature and present with joint swelling and genu valgum. Additional reported manifestations include clinodactyly, spindle-shaped fingers, and pectus excavatum.
ORPHACODE:	166024
SYNONYMS:	Multiple epiphyseal dysplasia-macrocephaly-distinctive facies syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	KIF7
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- kinesin family member 7

Related Gene Panels

- Ciliopathy (120 genes) - UGent