DISEASE:
SHORT syndrome

NAME:	SHORT syndrome
DESCRIPTION:	A rare disorder characterized by multiple congenital anomalies. The name is a mneumonic for the common features observed in SHORT syndrome that include; short stature, hyperextensibility of joints, ocular depression, Rieger anomaly and teething delay. Other common manifestations of SHORT syndrome are mild intrauterine growth restriction, partial lipodystrophy, delayed bone age, hernias and a recognizable facial gestalt.
ORPHACODE:	3163
SYNONYMS:	Lipodystrophy-Rieger anomaly-diabetes syndrome Rieger anomaly-partial lipodystrophy syndrome
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	PIK3R1
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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

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Related Gene Panels

- Short Stature (46 genes) - IPG