Belgian Genetic Tests database

DISEASE:
Isolated Pierre Robin syndrome

NAME:	Isolated Pierre Robin syndrome
DESCRIPTION:	A rare, congenital head and neck malformation characterized by the association of retrognathia and glossoptosis, with or without cleft palate, and respiratory obstruction.
ORPHACODE:	718
SYNONYMS:	Isolated Pierre Robin sequence
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SOX9
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/334

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