
DISEASE:
Short stature due to partial GHR deficiency

NAME:	Short stature due to partial GHR deficiency
DESCRIPTION:	Short stature due to partial GHR deficiency is a rare, genetic, endocrine disease characterized by idiopathic short stature due to diminished GHR function (decreased ligand binding or reduced availability of receptor), thus resulting in partial insensitivity to growth hormone.
ORPHACODE:	314802
SYNONYMS:	Short stature due to partial growth hormone receptor deficiency
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>GHR</u>
CREATED:	13 May 2019 - 01:02
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- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)
- [Centrum Medische Genetica - UZ Antwerpen](#)

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