DISEASE:
Autosomal dominant hyperinsulinism due to SUR1 deficiency

NAME:	Autosomal dominant hyperinsulinism due to SUR1 deficiency
DESCRIPTION:	A form of congenital diazoxide-sensitive diffuse hyperinsulinism due to ABCC8 variants and characterized by hypoglycemic episodes that are usually mild, escaping detection during infancy, and usually have a good clinical response to diazoxide. The autosomal dominant hyperinsulinism usually has a milder phenotype when compared to that resulting from recessive potassium (K-ATP) channel mutations.
ORPHACODE:	276575
SYNONYMS:	Autosomal dominant hyperinsulinemic hypoglycemia due to SUR1 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ABCC8
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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Source URL: http://gentest.healthdata.be/disease/1260