DISEASE:
Autosomal dominant hyperinsulinism due to Kir6.2 deficiency

NAME:	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency
DESCRIPTION:	A form of diazoxide-sensitive diffuse hyperinsulinism (DHI) characterized by hypoglycemic epiosodes that are usually mild, escaping detection during infancy, and usually a good clinical response to diazoxide, (but some are diazoxide resistant). Autosomal dominant hyperinsulinism due to Kir6.2 deficiency usually has a milder phenotype when compared to that resulting from recessive K+ (K-ATP) channel mutations (Recessive forms of diazoxide-resistant hyperinsulinism).
ORPHACODE:	276580
SYNONYMS:	Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency Dominant KATP hyperinsulinism due to Kir6.2 deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KCNJ11
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