

**DISEASE:****Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency**

<b>NAME:</b>	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency
<b>DESCRIPTION:</b>	A rare, congenital, isolated hyperinsulinism disorder characterized by diazoxide unresponsive recurrent episodes of hyperinsulinemic hypoglycemia resulting from an excessive insulin secretion by the pancreatic beta-cells due to a mutation in the ABCC8 gene. Pancreatic involvement is focal and can be cured by a selective partial pancreatectomy. Hypoglycemia may lead to variable clinical manifestations, ranging from asymptomatic hypoglycemia revealed by routine blood glucose monitoring to macrosomia at birth, mild to moderate hepatomegaly and life-threatening hypoglycemic coma or status epilepticus, further leading to poor neurological outcome.
<b>ORPHACODE:</b>	276598
<b>SYNONYMS:</b>	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>
<b>ANALYTE(S):</b>	<a href="#">ABCC8</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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