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**DISEASE:**  
**Pyruvate carboxylase deficiency, infantile type**

<b>NAME:</b>	Pyruvate carboxylase deficiency, infantile type
<b>DESCRIPTION:</b>	Infantile pyruvate carboxylase (PC) deficiency (Type A) is a rare, severe form of PC deficiency characterized by infantile-onset, mild to moderate lactic acidemia, and a generally severe course.
<b>ORPHACODE:</b>	353308
<b>SYNONYMS:</b>	Pyruvate carboxylase deficiency type A
<b>XREF(S):</b>	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
<b>ANALYTE(S):</b>	<u>PC</u>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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