

**DISEASE:**  
**Propionic acidemia**

<b>NAME:</b>	Propionic acidemia
<b>DESCRIPTION:</b>	Propionic acidemia (PA) is an organic aciduria caused by the deficient activity of the propionyl Coenzyme A carboxylase and is characterized by life threatening episodes of metabolic decompensation, neurological dysfunction and that may be complicated by cardiomyopathy.
<b>ORPHACODE:</b>	35
<b>SYNOMYS:</b>	Ketotic hyperglycinemia Propionic aciduria Propionyl-CoA carboxylase deficiency
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">MeSH</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>
<b>ANALYTE(S):</b>	<a href="#">PCCA</a> <a href="#">PCCB</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- Ichthyosis (gene panel)

### Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

### Related Analytes

- propionyl-CoA carboxylase subunit alpha
- propionyl-CoA carboxylase subunit beta

### Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL

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