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**DISEASE:**  
**Autosomal recessive cutis laxa type 2B**

<b>NAME:</b>	Autosomal recessive cutis laxa type 2B
<b>DESCRIPTION:</b>	A rare, hereditary, developmental defect with connective tissue involvement characterized by cutis laxa of variable severity, in utero growth restriction, congenital hip dislocation and joint hyperlaxity, wrinkling of the skin, in particular the dorsum of hands and feet, and progeroid facial features. Hypotonia, developmental delay, and intellectual disability are common. In addition, cataracts, corneal clouding, wormian bones, lipodystrophy and osteopenia have been reported.
<b>ORPHACODE:</b>	357064
<b>SYNONYMS:</b>	ARCL2, progeroid type ARCL2B Autosomal recessive cutis laxa type 2, progeroid type
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>
<b>ANALYTE(S):</b>	<a href="#">PYCR1</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

- Cutis Laxa / Geroderma osteodysplasticum (gene panel)

### Related Laboratories

- Centrum Medische Genetica - UZ Gent

### Related Analytes

- pyrroline-5-carboxylate reductase 1

### Related Gene Panels

- Cutis Laxa / Geroderma osteodysplasticum - UGent

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