Clinical Features:
Rhizomelic Chondrodysplasia Punctata (RCDP) is a peroxisomal disorder characterized by disproportionately short stature primarily affecting the proximal parts of the extremities, congenital contractures, characteristic ocular involvement, severe intellectual disability and spasticity. Characteristic facial features include broad nasal bridge, epicanthus, high-arched palate, dysplastic external ears and micrognathia. RCDP presents in the neonatal period and most affected individuals die in the first decade of life, although milder forms of RCDP can present with variable growth and developmental delays and survival into adulthood (1).

Molecular Genetics:
Mutations in PEX7 [OMIM#601757] account for more than 90% of patients with RCDP1 [OMIM#215100] (2). The finding of a deficiency of plasmalogens in red blood cells, increased plasma concentration of phytanic acid, and normal plasma concentration of very long chain fatty acids is consistently identified in individuals with PEX7 mutations (1). PEX7 is one of sixteen PEX genes that are involved in mammalian peroxisome assembly.

Mutations in GNPAT [OMIM#222765] and AGPS [MIM#600121] account for less than 10% of patients with RCDP2 [OMIM#222765] and RCDP3 [OMIM#60012] respectively (2). PEX7 transports AGPS into the peroxisome, where AGPS and GNPAT partner on the luminal membrane surface.

Inheritance:
RCDP follows an autosomal recessive inheritance pattern. Therefore, parents of an affected child are most likely obligate carriers. Recurrence risk for carrier parents is 25%. The prevalence of RCDP1 is estimated to be less than 1 in 100,000.

Test methods:
Our RCDP panel includes full gene sequencing for the PEX7, GNPAT and AGPS genes. Testing includes sequence analysis of all 10 coding exons and intron/exon boundaries of PEX7, all 16 coding exons and intron/exon boundaries of GNPAT, and all 20 coding exons and intron/exon boundaries of AGPS by direct sequencing of amplification products in both the forward and reverse directions.

Rhizomelic Chondrodysplasia Punctata Sequencing Panel (PEX7, GNPAT and AGPS sequencing)

<table>
<thead>
<tr>
<th>Sample specifications:</th>
<th>3 to 10 cc of blood in a purple top (EDTA) tube</th>
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<tbody>
<tr>
<td>Cost:</td>
<td>$1500</td>
</tr>
<tr>
<td>CPT codes:</td>
<td>81407</td>
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<tr>
<td>Turn-around time:</td>
<td>4 weeks</td>
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</tbody>
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Results:
Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

For more information about our testing options, please visit our website at dnatesting.uchicago.edu or contact us at 773-834-0555.
References:


Committed to CUSTOMIZED DIAGNOSTICS, TRANSLATIONAL RESEARCH & YOUR PATIENTS’ NEEDS