Rhizomelic Chondrodysplasia Punctata: Sequencing Panel

**Test Code:** MM610  
**Turnaround time:** 6 weeks  
**CPT Codes:** 81479 x1

### Condition Description

Rhizomelic chondrodysplasia punctata (RCDP) is a group of peroxisomal disorders that affects many parts of the body. Characteristic features include disproportionate shortening of bones of the upper arms and thighs (rhizomelia), specific bone changes seen on X-ray (punctate calcifications in cartilage), congenital cataracts, growth deficiency, distinctive facial features, and severe intellectual disability and developmental delay. Additionally, most children with this condition have respiratory tract infections, seizures, and may not live past the first decade of life. Some individuals may present with less severe symptoms that constitute a milder form of RCDP.

There are three different types of Rhizomelic Chondrodysplasia Punctata; type one is the most common. Types one, two, and three are caused by mutations in the PEX7, GNPAT (DHAPAT), and AGPS genes, respectively. RCDP is inherited in an autosomal recessive pattern and is estimated to affect less than 1 in 100,000 people.

### References:

3. OMIM (Type 1): [http://www.omim.org/entry/215100](http://www.omim.org/entry/215100)
   OMIM (Type 2): [http://www.omim.org/entry/222765](http://www.omim.org/entry/222765)
   OMIM (Type 3): [http://www.omim.org/entry/600121](http://www.omim.org/entry/600121)
   OMIM (PEX7): [http://www.omim.org/entry/601757](http://www.omim.org/entry/601757)

### Genes

AGPS, GNPAT, PEX7

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Rhizomelic Chondrodysplasia Punctata.

### Methodology

**Next Generation Sequencing:** In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

### Detection

**Next Generation Sequencing:** Clinical Sensitivity: Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

### Specimen Requirements

**Type:** DNA, Isolated

**Specimen Requirements:**
- Microtainer
- 3µg
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

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Specimen Collection and Shipping:
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Type: Whole Blood (EDTA)**

**Specimen Requirements:**
EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old): 3-5 ml
Older Children & Adults: 5-10 ml
 Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Related Tests**
- Individual sequencing is available for PEX7, GNPAT, and AGPS.