Renpenning Syndrome 1: **PQBP1** Gene Deletion/Duplication

**Test Code:** YQ  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81404 x1

### Condition Description

**Mutations**  
in the **PQBP1** gene (Xp11.23) cause  
X-linked recessive mental retardation that is often syndromic but can be  
nonsyndromic. **PQBP1** mutations have  
been associated with Renpenning syndrome, Sutherland-Hann syndrome, cerebropalatocardiac  
(Hamel) syndrome, and Golabi-Ito-Hall syndrome. Common features of X-linked  
mental retardation caused by **PQBP1** mutation seem to be mental retardation, microcephaly, and short stature. Considerable  
phenotypic variability is observed between families with different mutations  
even between families with the same mutation.

In addition to mental retardation, microcephaly, and short stature, other reported  
features include small testes, ocular colobomas, cardiac malformations, cleft  
palate, spastic diplegia, and anol anomalies. Facial characteristics include  
narrow and tall craniofacies with upslanting palpebral fissures, abnormal nasal  
configuration, cupped ears, and short philtrum. The nose may appear long or  
bulbous, with overhanging columella.

Historically, Renpenning syndrome  
has been associated with mental retardation with short stature, moderate  
microcephaly, but no remarkable facies and no other neurologic abnormalities.  
Sutherland-Haan syndrome has been associated with mental retardation, short  
stature, microcephaly, brachycephaly, spastic diplegia, small testes, and  
possibly intrauterine growth retardation. Cerebropalatocardiac (Hamel) syndrome  
has been associated with severe mental retardation with congenital heart defects,  
microcephaly, spasticity, short stature, cleft or highly arched palate, and  
other craniofacial abnormalities. Golabi-Ito-Hall syndrome has been associated  
with mental retardation, microcephaly, postnatal growth deficiency, and other  
anomalies, including atrial septal defect.

[Click here](#) for the OMIM summary on this condition.

### Genes

**PQBP1**

### Indications

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of Renpenning syndrome in individuals who have tested negative for sequence analysis  
- Carrier testing in adult females with a family history of Renpenning syndrome who have tested negative for sequence analysis

### Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes  
which cover the entire genomic region.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-  
target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic  
off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be  
interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

- *Preferred specimen type: Whole Blood*

**Type:** Whole Blood

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Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Special Instructions**

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of Emory Genetics Laboratory, please submit a copy of the sequencing report with the test requisition.

**Related Tests**

- Sequencing analysis of the PQBP1 gene is available (YP) and is required before deletion/duplication analysis.
- A CGH array-based test for deletion/duplication analysis of 64 different X-linked intellectual disability genes is available (OL).
- Prenatal testing is available to adult females who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.