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*Biochemical Genetics Laboratory*

*Cytogenetics Laboratory*

*Molecular Diagnostic Center*

## RETT SYNDROME

## MOLECULAR DIAGNOSTIC CENTER

Rett syndrome is a neurodevelopmental disorder that occurs in 1 in 10,000-15,000 girls as a result of mutations in the MeCP2 gene located at chromosome Xq28. Rett syndrome also occurs in boys at a lower frequency.

### REASONS FOR REFERRAL

- Confirmation of clinical diagnosis in patients with classical Rett syndrome
- Diagnostic testing in patients with variants of Rett syndrome
- Testing of family members of Rett syndrome patients or those who have non-syndromic mental retardation or autistic spectrum disorder
- Prenatal diagnosis

### TESTING METHODOLOGY

**Mutation scanning:** Our laboratory uses PCR followed by denaturing high performance liquid chromatography (dHPLC) to screen the entire MeCP2 gene for heteroduplex DNA, indicating a DNA sequence change. Samples containing a change are directly sequenced to determine the presence of a disease causing mutation or a benign single nucleotide polymorphism.

#### *Mutation Analysis*

80% detection rate in females with classical Rett syndrome

Test reporting follows the ACMG guidelines.

### SPECIMEN REQUIREMENTS

**Blood:** ACD (solution A or B) tubes:

*Adult: 5 mL*

*Child: 5 mL*

*Infant: 2-3 mL*

Requisition form must accompany specimen including ethnicity, clinical and family history information. Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact our laboratory.

**TURNAROUND TIME: 2-3 weeks**

**CPT CODES: 83891, 83898x6, 83903x6, 83904, 83912**

Rett info 5/04