Cincinnati Children's

Division of Human Genetics

MECP2-Related Disorders

Molecular Genetics Laboratory
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Additional information and test requisitions are available at:
www.cincinnatichildrens.org/molecular-genetics

Helping you fit the pieces together

Shipping Instructions

Please enclose test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:
Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

- Rett syndrome and its variants
- *MECP2*-related severe neonatal encephalopathy
- X-linked mental retardation

Mutations in *MECP2* result in broad phenotypic variability. In females, *MECP2* mutations may present as classic Rett syndrome characterized by a period of normal development followed by rapid regression of language and motor skills by two years of age. Atypical variants of Rett syndrome have also been described in individuals with *MECP2* mutations including mental retardation and autism spectrum disorders. Finally, mutations in *MECP2* have been identified in intellectually normal and mildly impaired women, presumably due to non-random X-inactivation.

In males, *MECP2* mutations classically present with severe neonatal encephalopathy with early lethality. A few males have been reported with typical Rett syndrome and *MECP2* mutations. Mutations in *MECP2* have also been described in association with X-linked mental retardation (PPMX) in some families.

INDICATIONS:

- Diagnostic testing in females with suspected Rett syndrome or variant
- Diagnostic testing in males with severe neonatal encephalopathy
- Diagnostic testing in selected individuals who had defied characterization including some patients with autism, Angelman syndrome and intellectual disability, among others.
- Prenatal diagnosis in families with an identified *MECP2* mutation
- Carrier testing in relative of a patient with a *MECP2*-related disorder

METHODOLOGY:

PCR-based sequencing of the coding regions and their exon/intron boundaries of the *MECP2* gene.

CLINICAL SENSITIVITY:

PCR-based sequencing of the coding regions and their exon/intron boundaries of the *MECP2* gene detects ~ 80% patients with classic Rett syndrome. Multiple exon deletions are identified in approximately 15-20% of patients and can be identified by multiplex ligation (MLPA), which is not offered by our laboratory. Mutations in *CDKL5* are also associated with Rett syndrome in a small minority of affected individuals. Both MLPA an *CDK5* analysis are clinically available.

ANALYTICAL SENSITIVITY:

The sensitivity of PCR-based DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed.
Mutations in regulatory regions or other untranslated regions are not detected by this test.
Multiple exon deletions, large insertions, genetic recombinational events and rare, primer site mutations are not be identified by this methodology.

SPECIMEN:

At least 3mLs of whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Phlebotomist must initial tube to verify patient's identity.

TURN-AROUND TIME:

3-4 weeks

COST:

Please call 1-866-450-4198 for institutional pricing or with any billing questions.

CPT CODES:

MECP2 sequence analysis:	81302
Family specific analysis:	81403

RESULTS:

Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Updated 12/2011