MECP2-Related Disorders

• 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

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

30 days

MECP2 sequence analysis:  81302
Family specific analysis:  81403

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.

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