London Health Sciences Centre
St JOSEPH'S HEALTH CARE LONDON
Pathology and Laboratory Medicine

RETT SYNDROME

Orderable – E-order/Requisition

Turnaround Time: 3 months

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
DNA Sequence Analysis

Test Schedule:
As required,
Monday to Friday 0800-1600 hours

Alternate Name(s):

MECP2

Specimen:

Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

Interpretive Comments:

Rett syndrome, represents one of the leading causes of mental retardation and developmental regression in girls. The majority of cases of sporadic Rett syndrome are caused by mutations in the gene encoding methyl-CpG-binding protein 2 (MeCP2). The MeCP2 protein binds methylated DNA and appears to regulate gene expression and chromatin structure. Genotype/phenotype analysis reveals that the phenotypic spectrum of MeCP2 mutations in humans in broader than initally suspected, and mutations have been discovered in Rett syndrome variants, mentally retarded males, and autistic children[1]. The MeCP2 protein is coded for by three exons which can be analysed by direct sequence analysis of PCR-amplified genomic DNA derived from peripheral blood leukocytes, the larger exon #3 being analysed in three overlapping fragments.

Reference

1. *Neul J. and Zoghbi H. Rett Syndrome: A prototypical neurodevelopmental disorder. The Neuroscientist. (2004) 10:118-128*

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Storage and Shipment:

Must be received in testing laboratory within 5 days of collection, shipped at room temperature by courier/overnight delivery.