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CLIA #: 14D0917593 CAP #: 18827-49

ARX Gene Sequencing

Clinical Features and Molecular Genetics:

Mutations in the *ARX* gene have been identified in patients with X-linked lissencephaly with ambiguous genitalia (XLAG) (1), West syndrome/cryptogenic infantile spasms (2), and X-linked mental retardation (MRX) (3). Preliminary findings and ongoing studies suggest that about 100% of patients with a clinical diagnosis of XLAG have mutations in the *ARX* gene (1) while up to 10% of patients with a clinical diagnosis of West syndrome/cryptogenic infantile spasms and MRX may have mutations in the *ARX* gene (2, 3).

Test methods:

We offer mutation analysis of all coding exons and intron/exon boundaries of *ARX* by direct sequencing of amplification products in both the forward and reverse directions. We also offer deletion/duplication analysis of the *ARX* gene by MLPA or oligonucleotide array-CGH to identify deletions/duplications of one or more exons. Partial exonic copy number changes and rearrangements of less than 400 bp may not be detected by array-CGH. Array-CGH will not detect low-level mosaicism, balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype. The sensitivity of this assay may be reduced when DNA is extracted by an outside laboratory. For best results, please provide a fresh blood sample for this testing.

ARX sequencing analysis

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	$900
CPT codes:	81404
Turn-around time:	4 weeks

ARX deletion/duplication analysis

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	$1000
CPT codes:	81403
Turn-around time:	4 weeks

Results:

Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

References:

1. Kitamura K, Yanazawa M, Sugiyama N et al. Mutation of ARX causes abnormal development of forebrain and testes in mice and X-linked lissencephaly with abnormal genitalia in humans. Nat Genet 2002: 32: 359-369.
2. Strømme P, Mangelsdorf ME, Shaw MA et al. Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. Nat Genet 2002: 30: 441-445.
3. Bienvenu T, Poirier K, Friocourt G et al. ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. Hum Mol Genet 2002: 11: 981-991.