

The University of Chicago Genetic Services



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ARX Sequencing for XLAG, West syndrome & X-linked MR

Clinical Features and Molecular Genetics:

Mutations in the *ARX* gene have recently 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. Sample submission paperwork and instructions are included with this packet.

Mutation analysis (sequencing)

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	\$1000
CPT codes:	83891, 83898 x2, 83904 x4, 83912
Turn-around time:	4 - 6 weeks

Testing for a known mutation in additional family members

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	\$390
CPT codes:	83891, 83898 x 2, 83894, 83912
Turn-around time:	3-4 weeks

Prenatal testing for a known mutation

Sample specifications:	2 T25 flasks of cultured cells from amniocentesis or CVS or 10 mL of amniotic fluid
Cost:	\$590
CPT codes:	83891, 83898 x 2, 83894, 83912, 99051
Turn-around time:	1-2 weeks

Results

You will be informed of the results of your case as soon as it has been completed. Results, along with an interpretive report, will be faxed and mailed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

Laboratory Faculty and Staff:

Soma Das, Ph.D.
Director, Molecular Genetics Laboratory
ABMG Certified Molecular Geneticist

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References:

1. Kitamura et al., Mutations of *ARX* cause abnormal development of forebrain and testes in mice, and X-linked lissencephaly with abnormal genitalia in humans. *Nature. Genet.* In Press
2. Stromme et al., Mutations in the human ortholog of *Aristaless* cause X-linked mental retardation and epilepsy. *Nature Genet.* 30:441-445, 2002
3. Bienvenu et al., *ARX*, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. *Hum. Mol. Genet.* 11:981-991, 2002