GATA4 MUTATION ANALYSIS JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY



GATA4 is a member of GATA family of zinc finger transcription factors. Members of this family recognize the GATA motif which is present in the promoters of many genes. *GATA4* gene contains 6 exons spanning 55 kb of genomic DNA that had been mapped to chromosome 8p23.1. Human *GATA4* gene encodes a protein of 442 amino acids. This protein is thought to regulate genes involved in embryogenesis and in myocardial differentiation and function. Multiple mutations in *GATA4* have been identified in patients with atrial septal defect type 2 (ASD2), atrioventricular septal defect 4 and ventricular septal defect 1. *GATA4* mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *GATA4* mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the *GATA4* gene. We strongly recommend initial testing of a clearly affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals.

REASONS FOR REFERRAL

Molecular confirmation of the diagnosis of congenital heart diseases, such as atrial septal defect type 2 (ASD2), atrioventricular septal defect 4 and ventricular septal defect 1.

METHODOLOGY

Genomic DNA is analyzed for *GATA4* mutations by automatic fluorescent DNA sequencing of the coding exons of the *GATA4* gene, as well as the exon/intron junctions and a portion of the 5' and 3' untranslated region. Patient DNA is sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members are analyzed only for the familial mutation by automatic fluorescent DNA sequencing.

SERVICE FEES			
	Direct and Institutional Billing	CPT Codes	
Index Case (Male or Female)	\$750 per sample	81405, G0452	
Additional Family Members	\$300 per sample; known familial mutation only	81403, G0452	

SENSITIVITY

DNA Sequencing Analysis: Approximately 99% detection of mutations in the coding exons of GATA4

SPECIMEN REQUIREMENTS

Blood (preferred): EDTA (purple-top) tubes: *Adult:* 5 cc *Child:* 5 cc *Tissue:* Frozen (preferred), RNA*later*, Formalin-fixed, Paraffin-embedded **Other Body Fluids:** Call to inquire

Infant: 2-3 cc

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