BAYLOR GENETICS

Whole Genome Sequencing with Short Tandem Repeats (STR) for a More Comprehensive Diagnosis

Disease	Gene	STR
Central hypoventilation syndrome, congenital, with or without Hirschsprung disease [MIM:209880]	*PHOX2B*	GCN
Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome [MIM:614575]	*RFC1*	AARRG
Corneal dystrophy, Fuchs endothelial, 3 [MIM:613267]	*TCF4*	CTG or CAG
Dentatorubral-pallidoluysian atrophy [MIM:125370]	*ATN1*	CAG
Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) [MIM:254800]	*CSTB*	CCCCGCCCCGCG
Fragile X syndrome [MIM:300624]; Fragile X tremor/ataxia syndrome [MIM:300623]; Premature ovarian failure 1 [MIM:311360] *	*FMR1*	CGG
Friedreich ataxia [MIM:229300] *	*FXN*	GAA
Frontotemporal dementia and/or amyotrophic lateral sclerosis 1 [MIM:105550]	*C9orf72*	GGGGCC
Global developmental delay, progressive ataxia, and elevated glutamine [MIM:618412]	*GLS*	GCA
Huntington disease [MIM:143100] *	*HTT*	CAG
Huntington disease-like 2 [MIM:606438]	*JPH3*	CTG
Intellectual developmental disorder, X-linked 109 [MIM:309548]	*AFF2*	CCG
Intellectual Disability, FRA12A type [MIM:136630]	*DIP2B*	CGG
Machado-Joseph disease [MIM:109150]	*ATXN3*	CAG
Myotonic dystrophy 1 [MIM:160900] *	*DMPK*	CTG
Myotonic dystrophy 2 [MIM:602668]	*CNBP*	CCTG
Neuronal intranuclear inclusion disease [MIM:603472]; Tremor, hereditary essential, 6 [MIM:618866]	*NOTCH2NLC*	GGC
Oculopharyngeal muscular dystrophy [MIM:164300]	*PABPN1*	GCN
Spinal and bulbar muscular atrophy of Kennedy [MIM:313200] *	*AR*	CAG
Spinocerebellar ataxia 1 [MIM:164400]	*ATXN1*	CAG
Spinocerebellar ataxia 2 [MIM:183090]	*ATXN2*	CAG
Spinocerebellar ataxia 6 [MIM:183086]	*CACNA1A*	CAG
Spinocerebellar ataxia 7 [MIM:164500]	*ATXN7*	CAG
Spinocerebellar ataxia 8 [MIM:608768]	*ATXN8OS*	CTA/G
Spinocerebellar ataxia 10 [MIM:603516]	*ATXN10*	ATTCT
Spinocerebellar ataxia 12 [MIM:604326]	*PPP2R2B*	CAG
Spinocerebellar ataxia 17 [MIM:607136]	*TBP*	CAG or CAA
Spinocerebellar ataxia 27B [MIM:620174]	*FGF14*	GAA
Spinocerebellar ataxia 36 [MIM:614153]	*NOP56*	GGCCTG

** Can be ordered as a standalone test.*

Improving Lives Through Personalized Care And Genetic Expertise

We provide diagnostic answers that matter - empowering patients and families to make informed decisions about their health and helping healthcare providers best manage care for their patients.

Whole Genome Sequencing — the Most Comprehensive Genetic Testing Solution

WGS assesses 98% of the genome, detecting known and potential disease-causing variants in both the protein-coding exons and clinically significant non-coding regions of the genome.

Analysis

- Genome-wide detection of single nucleotide variants (SNVs), insertions and deletions (indels), copy number variants (CNVs), structural variants (SVs), and absence of heterozygosity (AOH)
- Detection of mitochondrial DNA (mtDNA) variants
- Short tandem repeat (STR) detection in 29 genes
- Uniparental disomy (UPD) reported when both parents tested

WGS Offers

- **RNAseq[†] for enhanced variant classification**
- Variant interpretation aided by concurrent, familial comparator analysis
- Rapid testing options with written results starting at 5 days
- Flexible sample types accepted

Baylor Genetics: Making Genetic Testing More Accessible

- Online portal and Epic Aura for streamlined test ordering, supported by genetic counselors
- Billing support including prior authorization, verification of benefits, patient out-of-pocket cost estimation, and patient financial assistance program

To view our comprehensive test menu, visit **baylorgenetics.com**.

[†] *A qualified variant meets our prediction algorithm criteria (Splice AI) that RNAseq will provide additional functional information.*