	Baylor	GeneDx
Price	$7,000, no charge for expanded report if requested w/in 6 months of testing (includes all family members)	$7,000/trio, or $5,000 for proband and $2,500 for each additional family member past trio.
Turn-around Time	15 weeks (more commonly 10-12 weeks)	20-24 weeks
CPT codes	81400x1, 81401x1, 81402x1, 81403x1, 81404x1, 81405x1, 81406x1, 81407x1	81479x1
Sibling testing	- On a case by case basis (to be reviewed with lab GC before case sent) the lab will use the sibling's sample for Sanger confirmation (akin to use of parents' samples). - If want full exome on both sibs it would require ordering 2 exomes.	- When a trio is sent, whole exome sequencing is always performed on each member of the trio. - Adding a sibling to the trio for full exome is an additional $2,500
Reporting format	Single report for proband	Single report for proband. *If multiple affected individuals, reports for each affected (additional fee)
Platform	Illumina platform for next-generation sequencing. The exome capture is performed with Nimblegen reagents using a HGSC custom-designed capture reagent called VCRome 2.1.	Agilent SureSelect v4 capture kit Sequenced using the Illumina HiSeq
Coverage	Mean coverage of the exome is 100 – 120X. 95% of the exome is covered at ≥20X coverage. 85% covered at ≥40X.	Mean coverage is 100X. 90-95% assessed at 10x coverage, >98% of the target region will be covered at a minimum of 1x.
Filtering	- Minor allele frequencies, mutation databases and disease specific databases. - GeneTest genes and genes in OMIM are given priority in reporting. - A team of ABMG certified molecular lab directors & medical directors will interpret results based on disease phenotype. - Parental results of potentially causative mutations may yield additional information useful in predicting clinical significance.	By inheritance pattern, phenotype, severity of sequence change, population frequency ("control" and "disease" databases), function (in pathways, etc)
Release of raw data	- Ordering provider must be on IRB approved research project that allows for exome result review - Paperwork must be filled out by ordering physician and family - Cloud based data transfer - Fees: no charge	- Ordering provider needs to fill out form, must be signed by family - Fees: BAM files $100, VCF file no charge

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Reporting	**Focused report contains:** - Deleterious mutations in genes related to the disease phenotype - Unclassified variants in genes related to the disease phenotype. - Three categories are optional in focused report (opt-out): 1)Carrier status of AR conditions for reproductive purposes, 2)Pharmacogenetic: VKORC1/CYP2C9 (warfarin) & CYP2C19 (Plavix) 3)Medically actionable mutations as defined by Baylor. **All reported variants are confirmed by a second platform (typically Sanger sequencing)** **Incidental findings:** Family can opt in or out for medically actionable, carrier status for AR conditions and pharmacogenetics results. Regarding medically actionable findings specifically, family can: - opt into ACMG list plus others determined by Baylor - opt into ACMG list only - opt out of all	- Mutations and VUS in genes definitely related to phenotype - Mutations and VUS in Genes Possibly/Probably related to phenotype - Mutations in genes with unknown relationship to phenotype (very selective) **All reported variants are confirmed by Sanger sequencing in a new DNA extraction.** **Incidental findings:** Family can opt in or out of all incidental findings.
Secondary Findings	**Expanded report** can be ordered separately. Will contain information about genes unrelated to the phenotype, such as: - Deleterious mutations and unclassified variants in genes unrelated to the disease phenotype - Deleterious mutations in genes with no currently known association with disease in humans. Will **NOT** be report: - Heterozygous unclassified variants associated with recessive disorders, unless a deleterious mutation or a 2nd unclassified variant in the same gene is also detected. - Will **NOT** report findings in genes causing adult onset dementia syndromes such as early onset Alzheimer, for which there is no treatment.	**Expanded report** can be ordered separately & will include: - Variants in HGMD genes listed as "associated with" or infer a pre-disposition to a variety of clinical conditions, including adult-onset diseases, incurable diseases, or diseases with reduced penetrance. - Carrier status for autosomal recessive conditions. This includes variants with a population carrier frequency <3% in the 1000 Genomes Database. The expanded report will **NOT** include: - Novel variants not listed in HGMD, including novel truncating variants. - Secondary findings for any relatives of the primary individual tested by exome sequencing.

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Reanalysis	Baylor keeps list of newly discovered genes (published) and will review cases every 6 months for changes in these new genes. If a positive result is found, it will be reported to the referring physician in an addendum. **Families can opt in or out.**	The ordering provider may request a re-analysis of the exome sequencing data generated at the time of testing one year after the test report was issued. The first re-analysis can be requested at no charge. Additional re-analyses on an annual basis can be requested for a fee.
Research inclusion	-Grant to find more causes of mendelian disorders - If family opts in to research, then child's clinical and exome data will be transferred - Do not yet have a data mining ability for outside groups to mine this clinical/exome information to ascertain patients for studies (not sure how to do while protecting child's information) - Families can opt to be contacted by IRB approved researchers and/or have physician contacted	No information
Required documents	Pedigree & clinical assessment/notes (requested, but not required), consent form	Clinical form, medical records, pedigree, consent form
Other tests included:		
Chromosomal microarray	SNP array is performed for each sample as a quality assurance and sample identification step in the process.	May be included, additional fee
Mitochondrial genome	Included (no additional fee) – blood sample preferred	Included, but for additional fee (XomeDxPlus)

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Additional Information	-Parental samples will not be subjected to whole exome sequencing, but will be tested by targeted Sanger sequencing to confirm mode of inheritance, de novo status, etc. for mutations and/or variants in genes that are highly likely to be causative of disease. Not all mutations or variants will be confirmed by Sanger sequencing; the laboratory will prioritize which mutations/ variants to confirm based on the likelihood of being diagnostic in the case." Data files are available to researchers who have an institutional review board (IRB) approved protocol to authorize obtaining WES data from our clinical lab for further analysis.	- For trio testing, exomes of all three individuals are generated & sequenced - Sanger sequencing in a new aliquot of DNA prepared from the submitted sample to confirm all findings on the proband that are reported