GENERATION BEYOND

alamya health

Research Long-Read genome sequencing

Alamya Health, in conjunction with our laboratory partners, offers the Generation Beyond test, an all-in-one test that includes: research grade long-read (LR) whole genome sequencing, mitochondrial genome sequencing, DNA methylation profiling, and long-read transcriptome sequencing (cDNA).

Generation Beyond is intended for individuals who have had prior inconclusive genetic testing, or individuals for whom such testing could provide information to help guide diagnosis and clinical management. It can also be used for clinical research participants requiring LR sequencing +/- LR transcriptome. Alamya Health provides research reports along with raw data files (with some limitations).

Turnaround times for test results depend on the specific requirements of each individual case.

Generation Beyond is not yet approved as a clinical test.

Sequencing

Using Oxford Nanopore technology, the Generation Beyond test detects single nucleotide variants (SNVs), small insertions and deletions (indels) in regions of the genome inaccessible by short-read technologies, short tandem repeat expansions, structural variants (insertions, inversions, and translocations), and copy number variants (CNVs).

DNA methylation profiling

Assessment of genomic DNA methylation will be used to guide interpretation of variants in genes that encode epigenetic regulators and that could be associated with the clinical findings. Our Generation Beyond test includes Infinium MethylationEPIC microarray testing to focus on genes with well-established epigenetic signatures (see table below) that may indicate the presence of pathogenic variants.

Conditions with methylation signatures currently detected by our Infinium MethylationEPIC microarray*

Intellectual developmental disorder with autism and macrocephaly: *CHD8* gene	CHARGE syndrome: *CHD7* gene	Rahman syndrome: *HIST1H1E* gene
Intellectual developmental disorder, autosomal dominant 7: *DYRK1A* gene	Au-Kline syndrome: *HNRNPK* gene	Koolen-de Vries syndrome: *KANSL1* gene
Kleefstra syndrome 1: *EHMT1* gene	Kleefstra syndrome 2: *KMT2C* gene	Kabuki syndrome type 1: *KMT2D* gene
Sotos syndrome: *NSD1* gene	Bohring-Opitz syndrome: *ASXL1* gene	Kabuki syndrome type 2: *KDM6A* gene
Nicolaides-Baraitser syndrome: *SMARCA2* gene	Shashi-Pena syndrome: *ASXL2* gene	Down syndrome: chromosome 21 trisomy
Floating-Harbor syndrome: *SRCAP* gene	Weaver syndrome: *EZH2* gene	Chromosome 16p11.2 deletion syndrome, AUTS14A: 16p11.2 deletion
Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities: *SRCAP-upstream*	Cohen Gibson syndrome: *EED* gene	Chromosome 7q11.23 duplication syndrome: 7q11.23 duplication
Coffin-Siris syndrome 1: *ARID1B* gene	Imagawa-Matsumoto syndrome: *SUZ12* gene	Williams-Beuren syndrome: 7q11.23 deletion
KBG syndrome: *ANKRD11* gene	Dystonia 28, childhood onset: *KMT2B* gene	DiGeorge syndrome: 22q11 deletion
Developmental disorder and epileptic encephalopathy 94: *CHD2* gene	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder: *CDK13* gene	

* This signature listing is updated quarterly upon determination of robust epigenetic signatures.

Transcriptome sequencing

The transcriptome sequencing uses cDNA derived from whole blood RNA and can be used to resolve variants of uncertain significance (VUS) in individuals who have received genome sequencing.

Ordering and costs

For testing requests and costs, please contact: orders@alamyahealth.com or call (628) 203-5690. Generation Beyond can also be ordered through our website.

Visit our website at alamyahealth.com for more information on our Generation Beyond test.