

ARUP GENOME AND EXOME SEQUENCING

	GENOME SEQUENCING		EXOME SEQUENCING
	Rapid Whole Genome Sequencing (3005935)	Whole Genome Sequencing (3016493)	Exome Sequencing (3016583)
Test type	RWGS	WGS	WES
TAT for final report	≤7 days	14-21 days	21-28 days
Sample type accepted	Whole blood	Whole blood	Whole blood
Required samples	Patient Both parents: submit as Rapid Whole Genome Sequencing, Familial Control (3005928) or Rapid Whole Genome Sequencing, Familial Control with Report (3005933)	Patient only	Patient only
Additional recommended samples	None	Parental comparators: submit Whole Genome Sequencing, Familial Control (3016497)	Parental comparators: submit Exome Sequencing, Familial Control (3016589)
Methodology	NGS	NGS	NGS
Reported primary findings	Variant(s) potentially related to the patient's phenotype	Variant(s) potentially related to the patient's phenotype	Variant(s) potentially related to the patient's phenotype De novo and rare compound heterozygous variants in genes of unknown significance (only when overall result is nondiagnostic)
Secondary findings ^a	Reported for patient and parental comparators if opted in (additional fee applied for comparators)	Reported for patient and parental comparators if opted in (additional fee applied for comparators)	Reported for patient and parental comparators if opted in (additional fee applied for comparators)
Reanalysis	Available for a fee; order Whole Genome Reanalysis (3005939) TAT ≤21 days Secondary findings reanalyzed for patient only	Available for a fee; order Whole Genome Reanalysis (3005939) TAT ≤21 days Secondary findings reanalyzed for patient only	First reanalysis performed free of charge; order Exome Reanalysis (3001457) TAT 14-21 days Secondary findings reanalyzed for patient only

^aACMG gene list or other medically actionable variants

ACMG, American College of Medical Genetics and Genomics; CNV, copy number variant; mtDNA, mitochondrial DNA; NGS, next generation sequencing; RWGS, rapid whole genome sequencing; TAT, turnaround time; WES, whole exome sequencing; WGS, whole genome sequencing