Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/26/2002
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

MODY and Neonatal Diabetes Panel, Sequencing

ARUP test code 3001593

Monogenic Diabetes Specimen	Whole Blood

Monogenic Diabetes Interp: Negative

RESULT
No pathogenic variants were detected in any of the genes tested.

INTERPRETATION
No pathogenic variants were detected in any of the genes tested. This result decreases the likelihood of, but does not exclude, a heritable form of maturity-onset diabetes of the young (MODY) or neonatal diabetes. Please refer to the background information included in this report for a list of the genes analyzed, methodology, and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. If this individual has a family history, determination of a causative familial variant in an affected family member is necessary for optimal interpretation of this negative result. Further testing may be warranted if there is a familial variant that is not detectable by this assay. Genetic consultation is recommended.

COMMENTS
Likely benign and benign variants are not reported.
Variants in the following region(s) may not be detected by NGS with sufficient confidence in this sample due to technical limitations: None

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: MODY and Neonatal Diabetes Panel, Sequencing
CHARACTERISTICS: Maturity-onset diabetes of the young (MODY) is a group of inherited disorders that cause nonautoimmune diabetes mellitus with a typical onset before age 35. Most affected individuals have features that are atypical for type 1 and type 2 diabetes, including a lack of pancreatic islet autoantibodies, normal weight, triglycerides, and HDL, no acanthosis nigricans, low insulin requirements, and no ketoacidosis when insulin is omitted from treatment. Individuals with neonatal diabetes (ND) mellitus have complete or partial insulin deficiency and develop hyperglycemia by 6 months of age. Affected individuals often have intrauterine growth restriction, glucosuria, osmotic polyuria, severe dehydration, and failure to thrive.

EPIDEMIOLOGY: MODY accounts for 1-3 percent of all cases of

diabetes with no ethnic predilection; prevalence of ND is 1 in 160,000 in Austria and 1 in 215,000 in Slovakia.

CAUSE: Pathogenic germline variants in numerous genes.

INHERITANCE: Autosomal dominant or autosomal recessive, depending on the causative gene.

CLINICAL SENSITIVITY: Greater than 70 percent for MODY and greater than 73 percent for ND.

GENES TESTED: ABCC8*; APPL1; CEL*; EIF2AK3; FOXP3; GATA4; GATA6; GCK; HNF1A; HNF1B; HNF4A; INS; KCNJ11; NEUROD1; NEUROG3; PDX1; RFX6; SLC19A2; WFS1; ZFP57
*One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the targeted genes followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants that do not meet acceptable quality metrics. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes.

LIMITATIONS: A negative result does not exclude a heritable form of MODY or ND mellitus. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants, deep intronic variants, and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) variants, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.
SNVs and indels will not be called in the following regions due to technical limitations of the assay:
CEL (NM_001807) exons 1, 8, 9, 11
ABCC8 (NM_001351295) partial exon 14 (Chr11:17449973-17450018)

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Monogenic Diabetes Specimen	24-304-120972	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Monogenic Diabetes Interp	24-304-120972	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical