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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/2018
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Noonan Syndrome (PTPN11) Sequencing

ARUP test code 0051805

PTPN11 FGS Specimen	Whole Blood
Noonan Syndrome (PTPN11) Sequencing	Negative BACKGROUND INFORMATION: Noonan Syndrome (PTPN11) Sequencing CHARACTERISTICS OF NOONAN SYNDROME (NS): Short stature, developmental delay, dysmorphic facial features, congenital heart disease, broad or webbed neck, superior pectus carinatum and inferior pectus excavatum, low set nipples, cryptorchidism, coagulation and lymphatic disorders. CHARACTERISTICS OF LEOPARD SYNDROME: Lentigines, ECG abnormalities, ocular hypertelorism, pulmonary stenosis, abnormal genitalia, retardation of growth, and deafness. INCIDENCE: 1 in 1000 to 1 in 2500 for NS; rare for LEOPARD syndrome. INHERITANCE: Autosomal dominant. PENETRANCE: Unknown. CAUSE OF NS: Deleterious mutations in PTPN11, SOS1, RAF1, KRAS and other unidentified genes. CAUSE OF LEOPARD SYNDROME: Mutations in PTPN11 exons 7 and 12 as well as other unidentified genes. GENE TESTED: PTPN11. CLINICAL SENSITIVITY; 50 percent of NS is due to PTPN11 mutations; unknown for LEOPARD syndrome. METHODOLOGY: Bidirectional sequencing of the entire PTPN11 coding region and intron-exon boundaries. ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent. LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations and large deletions/duplications will not be detected. Mutations in genes, other than PTPN11, will not be detected. This assay is not designed to detect somatic variants associated with malignancy. Interpretation of this test result may be impacted if the patient has had an allogeneic stem cell transplantation. Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
PTPN11 FGS Specimen	19-261-105320	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Noonan Syndrome (PTPN11) Sequencing	19-261-105320	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