Patient Name	Centre
Age/Gender	OP/IP No/UHID
MaxID/Lab ID	Collection Date/Time
Ref By	Reporting Date/Time

SIN No:B2B5033162

TEST REQUESTED: **Mitochondrial DNA Sequencing**

METHOD USED

Next Generation Sequencing

CLINICAL INFORMATION/FAMILY HISTORY

Two month old male presented with double outlet right ventricle, ventricular septal defect, biventricular hypertrophy, he is being evaluated for related Genetic etiology.

TEST RESULTS

No clinically relevant mitochondrial variant has been detected

VARIANT INTERPRETATION

No significant variants were found with regards to the patient's symptoms.

SECONDARY FINDINGS

No pathogenic or likely pathogenic variants identified in the genes for which incidental findings are to be reported based on the ACMG-AMP guidelines.

Gene	Variant	Chromosomal Coordinates	Exon	Zygosity	Disease	Inheritance	Classification
NONE							

RECOMMENDATIONS

- Further investigations are recommended to assess the variants in nuclear genes, CNV and chromosomal aberrations.
- Genetic counselling is recommended.

Patient Name	Centre
Age/Gender	OP/IP No/UHID
MaxID/Lab ID	Collection Date/Time
Ref By	Reporting Date/Time

SIN No:B2B5033162

QUALITY THRESHOLD

Percentage (%) of Targeted Region Covered

Total data generated (Gb)	06.87
Reads aligned (%)	98.45
Q30 data (%)	95.51
Target region with 25X coverage	96.61
Target region with 50X coverage	86.13
Target region with 100X coverage	50.63

TEST DESCRIPTION

MitoDx is a complete mitochondrial DNA sequencing test. This test uses Next Generation Sequencing (NGS) technology to detect the cause of genetic disorders. It utilizes the patient's genotype information to detect the causative variants, preferentially. It assists the clinician in identifying the underlying cause of the disorder with certainty, solving the diagnostic odyssey for the patient. The clinician can personalize the treatment as per the clinical diagnosis of the patient.

TEST METHODOLOGY

Genomic DNA was extracted from the submitted specimen and targeted regions were sequenced using the Illumina sequencing platform system with average of 150bp paired-end reads. The DNA sequence was mapped to, and analyzed in comparison with, the published human genome build UCSC hg38 reference sequence. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage and data quality threshold values. The possible causative variants were prioritised based on the variant's predicted pathogenicity, frequency of occurrence in population and patient's phenotype with known disease causing genes from human and model organisms data. Analysis results are reported based on the recommendations of American College of Medical Genetics and Genomics and the Association for Molecular Pathology (ACMG-AMP), as described below:

Class 1	Pathogenic	This variant may directly contribute to the development of disease.
Class 2	Likely Pathogenic	There is a high likelihood that this variant is disease-causing. Additional evidence is expected to confirm this assertion of pathogenicity
Class 3	Variant of Uncertain Significance (VUS)	There is not enough scientific evidence at this time to support a more definitive classification of this variant.
Class 4	Likely Benign	As per current scientific evidence, this variant is not expected to have a major effect on disease. Additional evidence is expected to confirm this assertion. New evidence may demonstrate that this variant can contribute to disease.
Class 5	Benign	The variant does not cause disease.

In line with ACMG-AMP recommendations for reporting of secondary findings in clinical exome and genome sequencing, we report pathogenic variants and likely pathogenic variants only in the recommended genes for the recommended phenotypes.

LIMITATIONS

Inaccurate and/or incomplete clinical information might lead to misinterpretation of results. Only the variants located in genes that are potentially related to the proband's clinical phenotype are reported. Absence of a plausible explanation for the reported phenotype by exome sequencing does not exclude a genetic basis of the patient's condition. Intronic variants, repeat expansions, copy number variations or chromosomal rearrangements may not be reliably detected with exome Sequencing and therefore are not assessed. Due to technology limitations, certain genes may be poorly covered, impacting the test results. It is possible that the genomic region where a disease causing mutation exists in the proband was not captured using the current technologies and therefore was not detected. Additionally, it is possible that a particular genetic abnormality may not be recognized as the underlying cause of the genetic disorder due to incomplete scientific knowledge about the function of all genes in the human genome and the impact of variants in those genes. Only variants in genes associated with the medical condition, or thought to be clinically relevant potentially for the proband's medical condition, are reported here. Intronic variants, repeat expansions, copy number variations or chromosomal rearrangements can not be detected with mitochondrial sequencing. Due to technology limitations, large deletion and duplication variants cannot be con-fidently detected.

Patient Name	Centre
Age/Gender	OP/IP No/UHID
MaxID/Lab ID	Collection Date/Time
Ref By	Reporting Date/Time

SIN No:B2B5033162

DISCLAIMERS

This report provides information about the patient's mutations that may aid the physician's decision making process, but this test should not be the sole source of information for making decisions on patient care and treatment. The test should be interpreted in the context of standard clinical, laboratory, and pathological findings. Identification of a mutation in one or more of these genes does not guarantee activity of the drug in a given indication. Benign mutations and mutations in the intronic regions have not been included in this report.

The information provided in this report was collected from various sources that we believe to be reliable and quality control procedures have been put in place to ensure the information provided is as accurate, comprehensive, and as current as possible. The information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician. Patient care and treatment decisions should only be made by the physician after taking into account all relevant information available including but not limited to the patient's condition, family history, findings upon examination, results of other diagnostic tests, and the current standards of care. This report should only be used as an aid and the physician should employ clinical judgment in arriving at any decision for patient care or treatment.

(DR ATUL THATAI)
Director Molecular &
Cytogenomics

(DR NITIN DAYAL)
Prin. Cons. and Head
Hematopathology