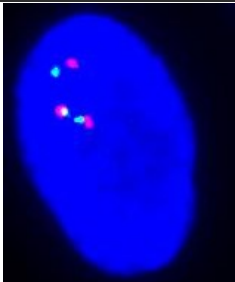


<b>Patient Name</b> : Mrs Dummy	<b>Sample Collected</b> : Aug 09, 2023, 03:23 PM.
<b>DOB/Age/Gender</b> : 41 Y 2 M 16 D/Female	<b>Report Date</b> : Aug 25, 2023, 03:33 PM.
<b>Patient ID / UHID</b> :XXX	<b>Barcode No</b> : XXX
<b>Referred By</b> : Dr. XXX	<b>Report Status</b> : Final Report
<b>Sample Type</b> : AMNIOTIC FLUID	

### DiGeorge Syndrome By FISH

**Clinical Indication** Anomaly scan shows ARSA present in the fetal heart, and rule out for digeorge syndrome (22q11.2) microdeletion.

**FISH Probe Used** DIGEORGE/VCFS TUPLE1 and 22q13.3 deletion Probe from CytoCell. The TUPLE 1 probe is 113 kb labelled in red and cover most of the TUPLE 1 (HIRA) gene at the chromosome loci 22q11.2 and NB5A3 (44 kb) probe labelled in green is located within 22q13.3 and covers the telomeric end of SHANK3 gene.

	<b>SPECTRUM GREEN (G)</b>	<b>SPECTRUM RED (R)</b>	<b>No. of Cells Analyzed</b>
	N85A3/SHANK3 gene (22q13.3)	TUPLE1/HIRA gene (22q11.2)	200

### N85A3/TUPLE1 :

Loci Analyzed	Signal Pattern	Normal Cut Off (%)	Percentage of Cells showing signal pattern	Result
N85A3/TUPLE1	2G/2R	>=2 %	100%	Negative


**ISCN Nomenclature** nuc ish (N85A3,TUPLE1)×2[200]  
(As per International System of Human Cytogenomic Nomenclature, ISCN, 2020)

**Interpretation** **NEGATIVE FOR DIGEORGE/VCFS SYNDROME, 22q11.2 microdeletion in all the analyzed interphase cells & metaphases.**

**Result** Fluorescence in-situ hybridization (FISH) analysis, revealed two red and two green signals (2R, 2G) in all the analyzed metaphases and interphase cells from the patient amniotic fluid sample thus showing normal FISH signal pattern for DIGEORGE/VCFS SYNDROME or negative for TUPLE1/HIRA gene 22q11.2 microdeletion.

(FISH is not intended for use as a standalone test for making clinical decisions)

\*\*\* End Of Report \*\*\*

  
**Dr. Ashish Fauzdar**  
 PhD (Genetics), AIIMS  
 Head of Clinical Genomics & Cytogenetics

Booking Centre :- Afmc(Ambady Scan Center-Cochin), .

Processing Lab :- Redcliffe Lifetech Pvt. Ltd., H-55, Sector-63, Noida, Uttar Pradesh - 201301

# Terms and Conditions of Reporting

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.
3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.
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**Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.**

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