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

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/31/1994
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome FISH, Metaphase

ARUP test code 2002299

Chromosome FISH, Metaphase

See Note (Ref Interval: Normal)

Test Performed: Chromosome FISH, Metaphase (CHR FISHM)
Specimen Type: Peripheral blood
Indication for Testing: Metaphase DiGeorge

RESULT
Abnormal FISH Result

22q11.2 (HIRA) Deletion: DETECTED

22q11.2 Deletion (DiGeorge/Velocardiofacial) syndrome

INTERPRETATION
This analysis showed a signal pattern consistent with deletion of the 22q11.2 (HIRA) locus. Structural abnormalities involving other loci, aneuploidy, and mosaicism have not been ruled out by this analysis.

Deletion of the 22q11.2 region is associated with DiGeorge/Velocardiofacial syndrome (DGS/VCFS). Clinical features in DGS/VCFS are variable and may include congenital heart defects (particularly conotruncal malformations), palatal abnormalities, characteristic facial features, developmental delay/intellectual disability, behavioral difficulties, immune deficiency due to absent or hypoplastic thymus, and hypocalcemia due to parathyroid hypoplasia (which may lead to seizures). Other findings may include feeding and swallowing problems, GI, renal, CNS, and skeletal anomalies, as well as hearing loss, growth hormone deficiency, autoimmune disorders, ophthalmologic abnormalities, enamel hypoplasia, autism, and psychiatric illness (particularly schizophrenia).

NOTE: The majority of 22q11.2 deletions involve recurrent breakpoints; however, genomic microarray analysis would be necessary to determine whether this finding represents a typical or atypical deletion.

Up to 10 percent of 22q11.2 deletions are inherited, sometimes from a mildly affected or unaffected parent. Parental FISH testing is recommended to evaluate the potential origin of this deletion and for recurrence risk counseling.

This assay only analyzes the DNA locus complimentary to the FISH probe for enumeration and localization of that sequence. This result does not rule out low-level mosaicism or copy number variants outside of the probe target.

This analysis was performed with the DiGeorge 22q11.2 TUPLE1 (HIRA)/22q13.3 (N85A3) probe (CytoCell). A total of 10 metaphase and 100 interphase cells were scored.

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

Recommendations:
1) Genetic counseling
2) Parental testing for the deletion by FISH analysis. This test is available, at a charge, through ARUP Laboratories. Please order test code 2002299, Chromosome FISH, Metaphase and request the DiGeorge probe.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

References:
1) McDonald-McGinn et al. 22q11.2 deletion syndrome. GeneReviews. 2020. (www.ncbi.nlm.nih.gov/books/NBK1523/). PMID: 20301696.
2) Burnside. 22q11.21 Deletion Syndromes: A review of proximal, central, and distal deletions and their associated features. Cytogenet Genome Res 2015;146(2):89 99. PMID: 26278718.
3) McDonald-McGinn and Sullivan. Chromosome 22q11.2 deletion syndrome
(DiGeorge syndrome/velocardiofacial syndrome). Medicine (Baltimore). 2011 Jan;90(1)1-18. PMID: 21200182.
4) Bassett et al. Practical guidelines for managing patients with 22q11.2 deletion syndrome. J Pediatr. 2011 Aug;159(2):332-9. PMID: 21570089.
5) Fung et al. Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genet Med. 2015 Aug;17(8):599-609. PMID: 25569435.
6) The International 22q11.2 Deletion Syndrome Foundation. (www.22q.org)
7) Unique: Understanding Rare Chromosome and Gene Disorders. (www.rarechromo.org)

Cytogenomic Nomenclature (ISCN):
ish del(22)(q11.2q11.2)(HIRA-).
nuc ish(HIRAx1,N85A3x2)

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis, Fish

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.

EER Chromosome FISH, Metaphase

See Note

Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome FISH, Metaphase	23-041-401731	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chromosome FISH, Metaphase	23-041-401731	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