Chromosome FISH, Metaphase

Patient: [redacted] DOB: [redacted] Age: 0 Gender: M **Patient Identifiers:** [redacted] **Visit Number (FIN):** [redacted]	Client: [redacted] Physician: [redacted]	ARUP Test Code: 2002299 Collection Date: 01/17/2019 Received in lab: 01/19/2019 Completion Date: 01/22/2019

Interpretation

Specimen Received
Specimen Type: Peripheral Blood
Reason for Referral: Tetralogy of Fallot
Test Performed: FISH, Metaphase

ABNORMAL FISH RESULT
22q11.2 (HIRA): deletion present

DIAGNOSTIC IMPRESSION:
Fluorescence in situ hybridization (FISH) analysis was performed with the VCFS TUPLE 1 probe (Cytocell).

This result is consistent with a clinical diagnosis of 22q11.2 deletion syndrome. Features associated with this disorder 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).

As up to 10 percent of 22q11.2 deletions are inherited from a mildly affected or unaffected individual, parental FISH testing is recommended.

Recommendations:
1) Genetic counseling
2) Parental FISH studies to determine if this deletion was inherited and to assess recurrence risk. This test is available, at a charge, through ARUP Laboratories. If ordering parental testing through ARUP Laboratories, please order test code 2002299, Chromosome FISH, Metaphase and request the DiGeorge FISH probe.

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

References and Resources:
1) McDonald-McGinn DM, et al. 2013. 22q11.2 deletion syndrome. GeneReviews. https://www.ncbi.nlm.nih.gov/books/NBK1523/. PMID: 20301696.
2) Burnside RD. 2015. 22q11.21 Deletion Syndromes: A review of proximal, central, and distal deletions and their associated features. Cytogenet Genome Res;146(2):89-99. PMID: 26278718.
3) McDonald-McGinn DM and Sullivan KE. 2011. Chromosome 22q11.2 deletion syndrome
(DiGeorge syndrome/velocardiofacial syndrome). Medicine

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(Baltimore). Jan;90(1)1-18. PMID: 21200182.
4) Bassett AS, et al. 2011. Practical guidelines for managing patients with 22q11.2 deletion syndrome. J Pediatr. Aug;159(2):332-9. PMID: 21570089.
5) Fung WL, et al. 2015. Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genet Med. Aug;17(8):599-609. PMID: 25569435.
6) The International 22q11.2 Deletion Syndrome Foundation. (www.22q.org)
7) The Unique Rare Chromosome Disorder Support Group. (www.rarechromo.org)

ISCN:
ish del(22)(q11.2q11.2)(HIRA-)

This result has been reviewed and approved by Erica F. Andersen, Ph.D., FACMG

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement A: aruplab.com/CS

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

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