Chromosome FISH, Metaphase

Patient: [redacted] DOB: [redacted] Age: [redacted] Gender: [redacted] **Patient Identifiers:** [redacted] **Visit Number (FIN):** [redacted]	Client: [redacted] Physician: [redacted]	ARUP Test Code: 2002299 Collection Date: 10/11/2016 Received in lab: 10/15/2016 Completion Date: 10/17/2016

Interpretation

Specimen Received
Specimen Type: Peripheral Blood (Slides)
Reason for Referral: DiGeorge Syndrome, Asymmetrical Cries Facies, IAA
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 analysis showed a deletion of this locus at 22q11.2 in 10 metaphase cells scored. This result is consistent with DiGeorge/ Velo-Cardio-Facial/ 22q11.2 deletion syndrome.

Clinical features associated with the DiGeorge/Velocardiofacial/22q11.2 deletion syndrome are variable and may include: conotruncal cardiac anomalies, palatal anomalies, immune dysfunction, hypocalcemia, developmental delay, learning disabilities, behavioral problems, characteristic facial features, and other minor features (see References).

The following resources may be useful for patient and family member education and support: The International 22q11.2 Foundation (www.22q.org/) and The Unique Rare Chromosome Disorder Support Group (www.rarechromo.org).

As up to 10% of these deletions are inherited, parental FISH testing may be considered (see Recommendations).

References:
1. Bassett et al. 2011. Practical guidelines for managing patients with 22q11.2 deletion syndrome. J Pediatr. 159(2):332-9.
2. Habel et al. 2012. Syndrome-specific growth charts for 22q11.2 deletion syndrome in Caucasian children. Am J Med Genet A. 158A(11):2665-71.
3. McDonald-McGinn DM, Sullivan KE. 2011. Chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). Medicine 90(1):1-18.
4. McDonald-McGinn et al. 2005. 22q11.2 Deletion Syndrome. GeneReviews. (http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=gr_22q11deletion)
5. Shaikh et al (2000) Chromosome 22-specific low copy repeats and the 22q11.2 deletion syndrome: genomic organization and deletion endpoint analysis. Hum Mol Genet. 9:489-501
6. Yagi et al (2003). Role of TBX1 in human del22q11.2 syndrome.

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Recommendations:
1. Genetic counseling
2. As some individuals with this 22q11.21 deletion might be mildly affected or apparently healthy, examination of parents by FISH may be considered to determine if a parent also carries this deletion and to assess recurrence risk. This test is available through ARUP Laboratories. There is a charge for this FISH testing. If ordering parental testing through ARUP Laboratories, please request test code 2002301, Array, Family Confirmation Study by FISH, and include the name/DOB of the proband.

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

This result has been reviewed and approved by , Ph.D., FACMG
Electronic Signature

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

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