

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: 01/31/2019
Received in lab: 02/02/2019
Completion Date: 02/07/2019

Interpretation

Specimen Received
Specimen Type: Peripheral Blood
Reason for Referral: Hypotonia, Developmental Delay (Motor),
Poor Feeding, Failure to Thrive
Test Performed: FISH, Metaphase

NORMAL FISH RESULT
22q11.2 (HIRA): deletion or duplication not detected

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

This analysis showed normal results with no evidence of a deletion or duplication of this locus at 22q11.2 in 10 metaphase cells scored.

FISH assays only analyze the DNA sequence representing the FISH probe for gain or loss of the sequence, and in the case of metaphase FISH, indicate the cytogenetic location of that sequence. A normal result does not rule out alterations elsewhere in the genome, small deletions or duplications within the sequence complementary to the probe, point mutations, or other mutational mechanisms yet unknown. Therefore, FISH should be considered as an adjunct to other genomic analyses.

ISCN:
ish 22q11.2(HIRAX2)
nuc ish 22q11.2(HIRAX2)

This result has been reviewed and approved by [REDACTED],
Ph.D., FACMG

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

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.

