

Patient ID SA00150304	Patient Name TESTINGRNV, DD22F-ABNORMAL	Birth Date 2012-01-15	Gender M	Age 10
Order Number SA00150304	Client Order Number SA00150304	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Jan 2022 08:49		

22q11.2 Deletion/Duplication, FISH

Result Summary

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Abnormal

Interpretation

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The result is abnormal. A deletion of HIRA was observed, consistent with a diagnosis of 22q11.2 deletion syndrome (Thomas, Clin Pediatr 36:253-66, 1997; Manji et al., Genet Med 3:65-66, 2001).

Approximately 7% of 22q11.2 deletions are inherited and there is significant phenotypic variability. Parental chromosome and FISH analysis could be performed to rule out a familial chromosome abnormality. A genetic consultation may be of benefit.

A chromosomal microarray study, test CMA CB (Chromosomal Microarray, Blood), is available to further define the extent of the region deleted and may also detect the presence of other relevant gains and losses of chromosomal material not visible by this FISH test. For more information regarding this test, please call 1-800-533-1710.

Result

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ish del(22)(q11.2q11.2)(HIRA-)
nuc ish(HIRAx1)

HIRA hybridized to only one chromosome 22 in ten metaphase cells and no HIRA signal was observed on any other chromosome.

Reason for Referral

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r/o 22q11.2 anomaly

Specimen

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Blood

Method

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FISH for 22q deletion or duplication syndromes using an Analyte Specific Reagent (ASR) probe for the critical region HIRA(TUPLE1) at 22q11.2 and a control locus (ARSA) at 22q13.3. Scoring method: Manual.

Disclaimer

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Analyte Specific Reagent (ASR). This test was developed using an analyte specific reagent. Its performance characteristics were determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the U.S. Food and Drug Administration. This FISH test does not rule out other chromosome abnormalities.

Released By

MCR

Ross A. Rowsey, Ph.D.

Received: 25 Jan 2022 08:55

Reported: 21 Feb 2022 07:32

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
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