

# FISH Analysis

**Patient Name:** Patient, Sample  
**Referring Physician:** Robert Jones, M.D.  
**Specimen #:** 10000000      **Client #:** 100000  
**Patient ID #:** 10000000 - 11111111 - 1 - FISH

Santa Fe Community Hospital  
5 Main Street  
Santa Fe, NM 87505

**DOB:** 03/11/98      **Date Collected:** 02/08/02  
**SSN:** \_\_\_-\_\_-\_\_      **Date Received:** 02/09/02  
                                 **Lab ID #:**  
                                 **Hospital ID #:**  
                                 **Specimen Type:** BLOOD

**Indications for Study:** Mental Retardation

## RESULTS: Abnormal Subtelomere FISH Results

### INTERPRETATION:

A deletion or rearrangement involving chromosome(s) \_\_\_\_ (and \_\_\_\_ ) was found by fluorescence in situ hybridization (FISH) utilizing 41 subtelomere probes (TelVysion<sup>®</sup>, Vysis, Inc.) that detect 43 subtelomeric regions (two ends for 1-12, 16-20, X and Y, and one end for 13-15 and 21-22). This screen cannot rule out unbalanced or balanced rearrangements of other chromosomal regions.

### DETAILS OF ADDITIONAL FISH WORK:

FISH utilizing subtelomeric probes has been shown to detect unbalanced rearrangements in 7% of individuals with moderate to severe mental retardation, with half of the cases having been inherited from a balanced carrier parent (Knight SJL, et al., Lancet 354: 1676-81, 1999).


It is recommended that subtelomere FISH analysis of blood from both parents be performed to determine if one of the parents carries a balanced form of this rearrangement (5cc blood in green-top sodium heparin tubes).

Five to ten cells were examined for each probe. The following probes were used:

1p (pVYS218C)	6q (pVYS229A)	11q (pVYS239A)	18p (pVYS249B)
1q (pVYS219A)	7p (pVYS230A)	12p (pVYS240C)	18q (pVYS250A)
2p (pVYS220B)	7q (pVYS231A)	12q (pVYS241A)	19p (pVYS251A)
2q (pVYS221B)	8p (pVYS232C)	13q (pVYS242C)	19q (pVYS252A)
3p (pVYS222A)	8q (pVYS233A)	14q (pVYS243B)	20p (pVYS253A)
3q (pVYS223B)	9p (pVYS234B)	15q (pVYS244B)	20q (pVYS254B)
4p (pVYS224A)	9q (pVYS235B)	16p (pVYS245B)	21q (pVYS255A)
4q (pVYS225A)	10p (pVYS236A)	16q (pVYS246A)	22q (pVYS207M)
5p (pVYS226C)	10q (pVYS237A)	17p (pVYS247B)	Xp/Yp (pVYS257A)
5q (pVYS227A)	11p (pVYS238A)	17q (pVYS248A)	Xq/Yq (pVYS258B)
6p (pVYS228B)			

This test was developed and its performance characteristics determined by Genzyme Genetics. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). Probes from Vysis, Inc. are considered analyte specific reagents (ASR s) by the FDA. These results should be used in conjunction with classical cytogenetic analysis and/or clinical information. All FISH probes undergo internal validation and quality control testing at Genzyme Genetics prior to use.

Signed:

  
Britt Ravnan, Ph. D.

Date: 03/06/02

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                                 **Lab ID #:**  
                                 **Hospital ID #:**  
                                 **Specimen Type:** BLOOD

**Indications for Study:** Mental Retardation

**RESULTS:** Normal Subtelomere FISH Results

## INTERPRETATION:

There is no evidence of a deletion or rearrangement by fluorescence in situ hybridization (FISH) utilizing 41 subtelomere probes (TelVysion®, Vysis, Inc.) that detect 43 subtelomeric regions (two ends for 1-12, 16-20, X and Y, and one end for 13-15 and 21-22). This screen cannot rule out unbalanced or balanced rearrangements of other chromosomal regions.


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