

Patient NAME : <b>Baby Dummy</b>	Report STATUS : Final Report
DOB/Age/Gender : 2 Y 2 M 24 D/Female	Barcode NO : XXX
Patient ID / UHID : XXX	Sample Type : Whole Blood Sod. Heparin
Referred BY : Dr. XXX	Report Date : Nov 22, 2024, 10:57 AM.
Sample Collected : Oct 25, 2024, 09:12 AM.	

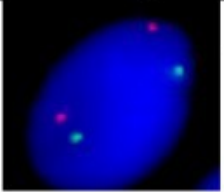
**Deletion 7Q By FISH**

CLINICAL DIAGNOSIS: KABUKI syndrome ?

FISH PROBE: Williams syndrome (Elastin gene) 7q11.23 region probe. Williams syndrome is the result of a commonly deleted region of 7q11.23 containing the Elastin (ELN) gene locus. This syndrome is believed to be contiguous gene syndrome. The probe consists of a probe for the elastin gene locus, the LIMK1 gene, and the D7S613 locus. The probe also contains a control probe for the region 7q36.3

METHOD: Fluorescence in situ hybridization (FISH)

NO. OF CELLS STUDIED: 20					
	ORANGE 7q11.23 (ELN)	GREEN 7q36.3 (Control)	NO. OF CELLS SHOWING DELETION	NO. OF CELLS NOT SHOWING DELETION	INTERPRETATION
Chromosome	7	7			
SIGNALS PER CELL	2	2	--	20	100% Normal
SIGNALS PER CELL	1	2	00	--	00% Positive

	INTERPRETATION
	Microdeletion of the region <b>7q11.23 not found</b> in all the cells studied.

**NOMENCLATURE:** nuc ish (ELNx2,7qTelx2) [20]

(As per International System of Human Cytogenetic Nomenclature, ISCN, 2020)

**RESULT: NEGATIVE** for Williams syndrome, 7q11.23 microdeletion in all the analyzed interphase cells & metaphases.

(FISH is not intended for use as a standalone test for making clinical decisions)

\*\*\* End Of Report \*\*\*



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Booking Centre :- Genomics Walk-In Redcliffe Labs- Noida, H55 Sec 63 Noida Uttar Pradesh  
Processing Lab :- Redcliffe Lifetech Pvt. Ltd., H-55, Sector-63, Noida, Uttar Pradesh - 201301

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