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Name: B/o Aswathy **MRD No:** 1784267

DOB: 20/12/2016 **Age:** 1 year

Referred by: Dr.Sheela Namboothiri **Sex:** Male

Nature of sample: Peripheral venous blood **HCL No:** N 064

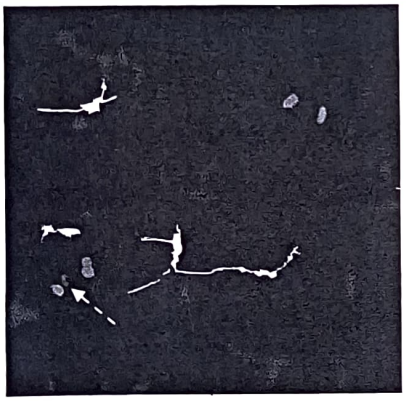
Investigation Requested: Angelman Syndrome

Sample Collected on: 17/05/18 **Reported on:** 23/05/18

Clinical History: Hand flapping, fair complexion

Method: Fluorescent *in situ* hybridization (FISH) was performed on metaphases of cultured lymphocytes using LSI SNRPN (Spectrum Orange), CEP 15(D15Z1) (Spectrum Aqua), LSI PML (Spectrum Green) Tri colour DNA Probe (Vysis).

Locus	Spectrum Aqua CEP (Control)	Spectrum Orange SNRPN	Spectrum Green PML	No. of metaphases analysed	Interpretation
	CEP	15q11.2	15q22-24		
Signal per metaphase	2	1	2	45	100% deletion



A metaphase and nucleus showing two aqua, two green and one orange signal indicating deletion.

Comments:

ISCN Result: ish del(15)(q11.2q12)(SNRPN-,D15S10-)

Interpretation

FISH analysis was performed with the 15q11.2-q12 probe (SNRPN, D15S10) and a PML control probe. This analysis showed a deletion of chromosome 15q11.2-q12 region in all metaphases and interphase nuclei scored. This result is consistent with the diagnosis of either Prader-Willi or Angelman syndrome, depending on parent-of-origin

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