LABORATORY REPORT

Redcliffe labs

MLPA Test

Patient		Sample		Clinician	
Name	DUMMY	**SampleType**	NA	**Name**	NA
Gender	NA	**Sample ID/ Specimen ID**	NA	**Hospital**	NA
DOB/ Age	NA	**Date and Time of Sample Collection**	DD-MM-YYYY	**Address**	NA
Place	NA	**Date and Time of Sample Received**	DD-MM-YYYY	**Indication**	XXY advised to get tested for Prader Willi Syndrome by MLPA.
Phone No. / Email ID	NA	**Date and Time of Sample Reported**	DD-MM-YYYY		

CLINICAL DIAGNOSIS/SYMPTOMS

NA

RESULT

- **No microdeletion and duplication is detected in Prader-Willi critical region(15q11.2-q13).**

Sr. No.	Gene	Location	Deletion/Duplication	Final Ratio (FR)	Reference
1	*TUBBGCP5*	Exon8	No deletion and duplication	0.99	0.80<FR<1.20
2	*MKRN3*	Exon1	No deletion and duplication	0.98	0.80<FR<1.20
3	*MAGEL2*	Exon1	No deletion and duplication	0.97	0.80<FR<1.20
4	*NDN*	Exon1	No deletion and duplication	0.96	0.80<FR<1.20
5	*SNRPN-region*	Exon1, U1b, Intr.u2, u5, E3, E7& Hb2	No deletion and duplication	~1	0.80<FR<1.20
6	*UBE3A*	Exon 2, 3, 4, 5 & 10	No deletion and duplication	~1	0.80<FR<1.20
7	*OCA2*	Exon 3 & 23	No deletion and duplication	~1	0.80<FR<1.20
10	*ATP10A*	Exon 1 & 15	No deletion and duplication	~1	0.80<FR<1.20
11	*GABRB3*	Exon 7 & 9	No deletion and duplication	~1	0.80<FR<1.20

- **Normal methylation pattern is observed in SNRPN region. Hence, No imprinting abnormalities is detected inSNRPN region as compared to Normal control sample.**

Sr. No.	Gene	Location	Methylation Status	Results	Final Ratio (FR)	Reference
1	*MAGEL2*	Exon1	50% Methylated	Normal	0.55	0.4≤FR≤0.65
2	*SNRPN-region*	Intr.2	50% Methylated	Normal	0.56	0.4≤FR≤0.65
3	*UBE3A*	Exon 1	Unmethylated	Normal	0	FR<0.05
4	*ESCO2* (Control)	Exon 1	Unmethylated	Normal	0	FR<0.05
5	*SLC9A2* (Control)	Exon 2	Unmethylated	Normal	0	FR<0.05

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All Lab results are subject to clinical interpretation by qualified medical professional and this report is not subject to use for any medico-legal purpose.

INTERPRETATION

No microdeletions and imprinting abnormalities is detected in the Prader-Willi critical region in Gargi Gupta. Sample from Gargi Gupta was referred to our laboratory for molecular testing for Prader Willi/Angelman Syndrome. Prader Willi/Angelman syndrome results by a deletion in the paternal chromosome 15 or by maternal uniparental disomy.

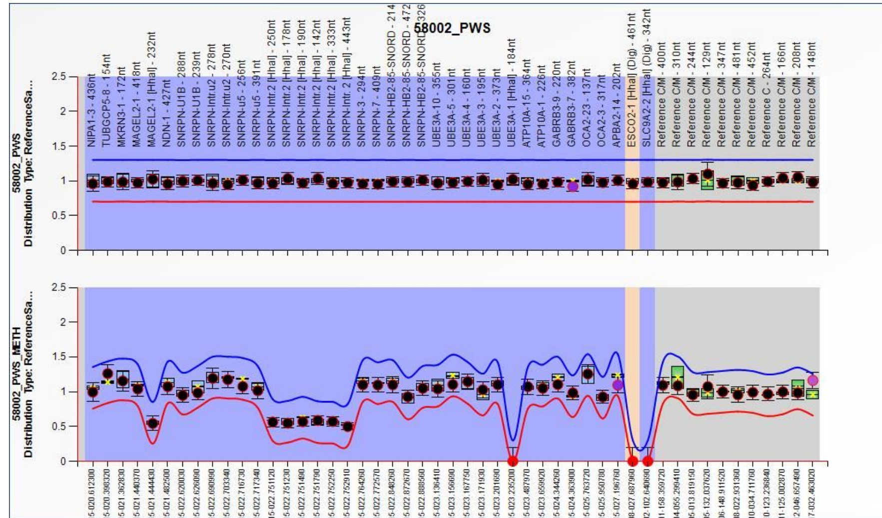

Fig-1: NA_NA_RATIO CHART: ME028 Probemix

Comment: The result must be interpreted in the context of the individaul's clinical and biochemical profile.

Genetic counselling is advised.

METHODOLOGY

Mutational analysis has been performed on genomic DNA by multiplex ligation probe dependent amplification (MLPA, MRC Holland) using SALSA MLPA probe mix ME028, SALSA Hha1, EKFAM kit for *PWS/AS* (OMIM#176270). Analysis was
done by Coffalyser (designed by MRC-Holland) .

DISCLAIMER

- Methylation variants and copy number variations (CNVs) discovered by many sequential ME028 Prader -Willi/Angelman probes should be verified using a separate method. Particularly, CNVs discovered by a single probe always need to be confirmed by an additional technique.
- The MLPA test will not detect the point mutations in the 15q11 region. It is therefore recommended to use MLPA in combination with sequence analysis. Thus, it is advised to employ this SALSA MLPA probemix in conjunction with a sequence analysis of the UBE3A gene to diagnose Angelman Syndrome.
- A point mutation or polymorphism in the sequence detected by a probe, which results in reduced probe binding efficiency, can also cause a reduction in relative peak area. Therefore, single exon deletions detected by MLPA should always be confirmed by other methods like multiplex PCR or sequencing.
- MLPA cannot detect any changes that lie outside the target sequence of the probes and will not detect most inversions or translocations. Even when MLPA did not detect any aberrations, the possibility remains that biological change in that gene or chromosomal region do exist but remain undetected.

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References:

- Cassidy, S. B., Schwartz, S., Miller, J. L., & Driscoll, D. J. (2012). Prader-willi syndrome. Genetics in medicine, 14(1), 10- 26.
- Khadilkar, V., Jagtap, V., Mathew, J., Phadke, N., Khatod, K., Kelkar-Ramanan, K., & Khadilkar, A. (2015). Genetic testing in Indian patients with Prader-Willi syndrome using methylation specific multiplex ligation dependent probe amplification (MS-MLPA). International Journal of Pediatric Endocrinology, 2015, 1-1.
- Driscoll, D. J., Miller, J. L., Schwartz, S., & Cassidy, S. B. (2017). Prader-willi syndrome. GeneReviews®[Internet].
- Henkhaus, R. S., Kim, S. J., Kimonis, V. E., Gold, J. A., Dykens, E. M., Driscoll, D. J., & Butler, M. G. (2012). Methylation- specific multiplex ligation-dependent probe amplification and identification of deletion genetic subtypes in Prader -Willi syndrome. Genetic testing and molecular biomarkers, 16(3), 178-186.
- https://support.mrcholland.com/downloads/files/1

#this test is not under NABL scope.

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

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DISCLAIMER

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