PROGENE LAB
For Medical And Genetic Tests
معمل بروجين للتحاليل الطبية

PRADER-WILLI (PW) SYNDROME

Methylation-specific MLPA (MS-MLPA)

Patient Name: **********	**اسم المريض:** **********
Sample type: Blood sample	**DOB:** **********
Patient phone No.: **********	**Sex:** Male
Referring Doctor: **********	**City, Country:** **********
Report type: Methylation Study for Prader Willi Syndrome	**Sampling date:** **********
Report date: **********	
Reason of referral: Suspicion of genetic etiology based on clinical features	

Clinical Information: 10- year- old male child born to non-consanguineous parents complaining of delayed mile stones, Polyphagia, ADHD and social communication disorder.

Summary of The Results

Negative result

NO deletions, duplications or alterations in methylation have been detected in the regions included in this analysis, associated with Prader Willi syndrome (OMIM#176270).

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Interpretation of the test results:

Prader Willi´s diagnosis in this patient is unlikely. This result is consistent with a normal dose and methylation pattern of the 15q11.2.

This study allows the detection of deletions and alterations in imprinting (parental unidisomy or alterations in methylation), responsible for 99% of Prader-Willi cases[1].

This report must be interpreted by a specialist within the clinical context and the patient's family history in conjunction with other laboratory findings.

Recommendation:

- Genetic counseling has to be offered to the family.

Methods:

- Extraction of DNA from peripheral blood from the sample received.
- MS-MLPA (Methylation Specific-Multiplex ligation probe amplification), Probemix ME028-D1-PWS/AS (MRC-Holland) containing probes that hybridize to genes TUBGCP5 (NM_019066.4), NIPA1 (NM_144599.4), MKRN3 (NM_005664.3), MAGEL2 (NM_019066.4), NDN (NM_002487.2), SNRPN (NM_022807.3), UBE3A (NM_130838.1), ATP10A (NM_024490.3), GABRB3 (NM_021912.4), OCA2 (N000275.2) and APBA2 (NM_005503.3) throughout the 15q11.2-13 region. The probes and reagents used in this assay are f experimental use.
- Analysis by capillary electrophoresis (Applied Biosystems) and Coffalyser software (MRC-Holland) which compares each sample wi controls semi-quantitatively.
- Detected deletions/imprinting defects are confirmed from a new DNA extraction if possible.
- In the case of detecting abnormal signals in a single probe, sanger sequencing of this region is performed.

Test restrictions and limitations:

- The MLPA technique does not allow the detection of point mutations not considered in the test itself, deletions or duplications that are outside the study region, inversions, translocations and chromosomal rearrangements.
- The presence of possible mosaicisms may not be detected by MLPA, which could potentially lead to false-positive or false-negative results. According to the vendor, tile identification is outside the detection limits of the MLPA technique. However, from the laboratory, those cases suggestive of mosaic will always be reported in order that these results are interpreted in the clinical context of the patient.

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- In cases of methylation defects, the probemix ME028 cannot discriminate between uniparental disomy and imprinting defects. For this, it would be necessary to study microsatellites in both the patient and the parents.
- Confirmation by another method is required when copy number changes are due to a single probe. There may be polymorphisms/mutations in the region that prevent probe binding.
- The genetic studies carried out in the SYNLAB laboratory are carried out following the quality recommendations of the international good practice guides to minimize the possibility of errors in any of the phases of the process.

References:

1- Driscoll DJ, Miller JL, Schwartz S, Cassidy SB. Prader-Willi Syndrome. 1998 Oct 6 [Last Revision: November 2, 2023]. GeneReviews.

With kind regards

Clinical Laboratory Geneticists Staff:

Prof. Dr. Marwa Elsharkawy

Associate Prof. Noha Abd Elhalim

Signature

Prof. Dr. Mohamed Abd Elmonem

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