آغا خان یونیورسٹی ہسپتال، کراچی
The Aga Khan University Hospital, Karachi

Detection of Prader-Willi/Angelman syndromes by Methylation Specific PCR

Department of Pathology and Laboratory Medicine

UPDATE NO. 11, VOL: XXIX, 2023

The information contained in this flyer is intended for healthcare professionals.

December 2023

What's New

Methylation specific PCR (MS-PCR) is a reliable diagnostic tool for Prader Willi Syndrome (PWS) and Angelman Syndrome (AS) patients. This test evaluates the methylation state of the SNRPN gene's CpG island and enables the rapid molecular diagnosis of PWS or AS.

Introduction:

The Prader-Willi (PWS) and Angelman (AS) syndromes are two separate intellectual disability disorders caused by chromosome 15q11-q13 deletion, uniparental disomy, or imprinting abnormalities. PWS neonates are hypotonic, have a faint cry, and are poor feeders, although they may improve with time. Seizures, inappropriate laughing, ataxic gait, puppet-like upper limb motions, lack of speech, and open mouth with protruding tongue are all symptoms of AS.

The PWS/AS region is localized on the proximal long arm of chromosome 15 (15q11.2-q13). It lies within a smaller 2.5 Mb differentially imprinted region. PWS/AS is a contiguous gene disorder; studies indicate that the complete phenotype is due to the loss of expression of several genes. It is also an example of an imprinted condition, as the expression of relevant genes in the 15q11.2-q13 region is dependent on parental origin.

Intent of Use:

MS-PCR is used for the detection of deletion in 15q11.2-q13 critical region in patients suspected of Prader Willi/Angelman syndrome. This test should be incorporated into the diagnostic work up for PWS or AS subjects particularly in those with normal molecular cytogenetic studies.

Important Note:

Test results should be interpreted in context of clinical findings, history and other laboratory data.

Specimen Requirement

4 ml Blood required in EDTA tube.

Principle:

MS-PCR is based on amplification of sodium bisulfite treated patient's DNA with primers specific to methylated and unmethylated region of SNRPN gene.

Charges:

Rs.19,450/ *Revisions may apply.

Schedule:

Test performed 1st and 3rd Monday; Report issued after 5 working days.

For more information please call: 021 3486 1620
or Email: laboratory@aku.edu

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