Cat Nr/REF: KBI-40109

English

For professional use only

Poseidon™ Repeat Free™ Prader-Willi SNRPN (15q11) & PML (15q24) Control probe

Introduction:

Prader-Willi syndrome (PWS) is a complex, multisystem disorder. The major clinical features include neonatal hypotonia, developmental delay, short stature, behavioral abnormalities, childhood-onset obesity, hypothalamic hypogonadism, and characteristic appearance. In approximately 70% of cases this is the result of deletion of this region from the paternal chromosome 15. These deletions are optimally detected by FISH utilizing a probe for the SNRPN (small nuclear ribonucleoprotein N) gene region. Angelman syndrome (AS) is a clinically distinct disorder from PWS that can be difficult to diagnose approximately 70% of cases of AS have a deletion of 15q11-q13 in the maternally contributed chromosome 15. In most cases, this is the same deletion as that identified in PWS, but the use of the more specific probe UBE3A is recommended for Angelman syndrome.
Both syndromes may also result from uniparental disomy which is not detectable by FISH analysis.

Intended use:

The PW/AS **SNRPN** region probe is optimized to detect copy numbers of the SNRPN gene region at 15q11.
The **PML** (promyelocytic leukemia) gene specific DNA probe at 15q24 is included as control probe.

The probe is recommended to be used in combination with a Poseidon FISH Kit providing necessary reagents to perform FISH (KBI-60002, KBI-60003 or KBI-60001) for optimal results.

Critical region 1 (red):

The **SNRPN** specific DNA probe is direct-labeled with Platinum*Bright*550.

Control region 2 (green):

The **PML** control DNA probe is direct-labeled with Platinum*Bright*495.

Reagent:

Poseidon probes are direct-labeled DNA probes provided in a ready-to-use format. Apply 10 µl of probe to a sample area of approximately 22 x 22 mm.

Please refer to the Instructions for Use for the entire Poseidon FISH protocol.

Poseidon Repeat Free probes do not contain Cot-1 DNA. Hybridization efficiency is therefore increased and background, due to unspecific binding, is highly reduced.

Interpretation:

The **SNRPN** probe is designed as a dual-color assay to detect deletions at 15q11. Deletions involving the SNRPN gene region will show one red signal and two green signals at the PML (15q24) control region (1R2G). Two single color red (R) and green (G) signals will identify the normal chromosomes 15 (2R2G)

	Normal Signal Pattern	Del 15(q11) SNRPN
Expected Signals	2R2G	1R2G

References:

Mutirangura A et al.Genomics. Dec;18(3):546-52, 1993
Trent RJ et al. J Med Genet. Sep;34(9):714-8.,1997
Christian SL et al, Genome Res. Feb;8(2):146-57, 1998

POSEIDON™
REPEAT-FREE™ FISH PROBES

Application Manual

KBI-40109
MD Prader-Willi SNRPN (15q11) / PML (15q24)

IVD
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CE

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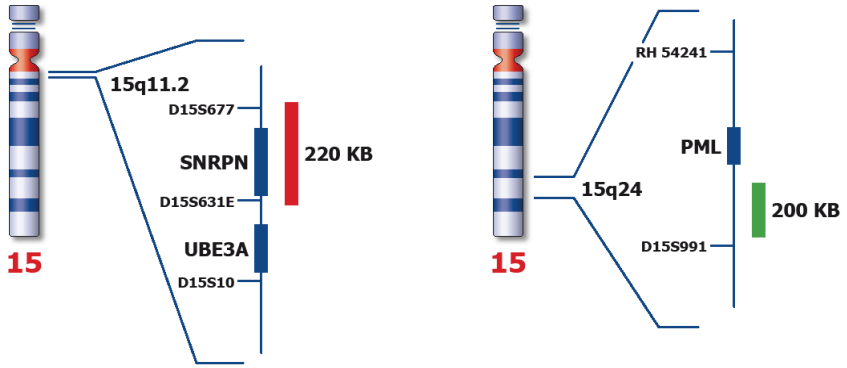

Not to scale

Application manual