Kreatech™ FISH probes

Product Information Sheet

KBI-45109

Prader-Willi SNRPN (15q11) / PML(15q24)

IVD

DANGER

FORMAMIDE

2°C 8°C

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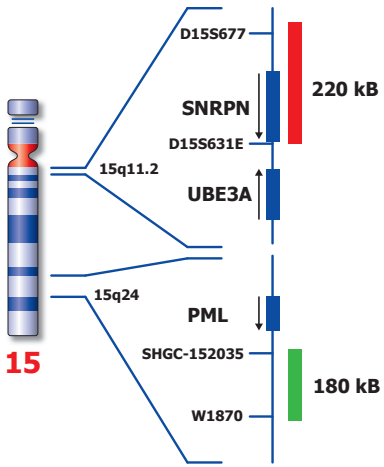

Not to scale

KBI-45109

Kreatech™ Prader-Willi SNRPN (15q11) / PML (15q24) FISH 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, previously known as PWCR, SMN, HCERN3) 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 **Prader-Willi SNRPN** region probe is optimized to detect copy numbers of the SNRPN gene region at 15q11. The **PML** (promyelocytic leukemia) gene specific FISH probe at 15q24 is included as control probe.

The probe is recommended to be used in combination with one of the Kreatech Pretreatment kits providing necessary reagents to perform FISH on various sample types for optimal results. (see also www.LeicaBiosystems.com and look for Kits & reagents)

Critical region 1 (red): The **Prader-Willi SNRPN** specific FISH probe is direct-labeled with Platinum*Bright*™550.
Control region 2 (green): The **PML** specific FISH probe is direct-labeled with Platinum*Bright*™495.

Reagent:

Kreatech 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 Kreatech FISH protocol.

Kreatech FISH probes are REPEAT-FREE™ and therefore do not contain Cot-1 DNA. Hybridization efficiency is increased and background, due to unspecific binding, is highly reduced.

Interpretation:

The **Prader-Willi SNRPN (15q11) / PML (15q24)** FISH 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(15q11) 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

Warning and precautions: In case of emergencies check SDS sheets for medical advice. SDS sheets may be obtained by either contacting Leica Technical Support or visiting www.LeicaBiosystems.com. DNA probes contain formamide which is a teratogen; do not inhale or allow skin contact. Wear gloves and a lab coat when handling DNA probes. All materials should be disposed of according to your institution's guidelines for hospital waste disposal.

Reagent Storage and Handling:

Store at 2-8 °C. Reagents should not be used after the expiration date on the vial label.

TECHNICAL SUPPORT

Technical support is available at www.LeicaBiosystems.com or +31 20 6919181 or via e-mail: kreatech-support@leicabiosystems.com.

CUSTOMER SERVICE

Kreatech probes may be ordered through Leica Customer Service +31 20 6919181 or order via e-mail: purchase.orders@leica-microsystems.com.