

Kreatech™ FISH probes

Product Information Sheet

KBI-40110

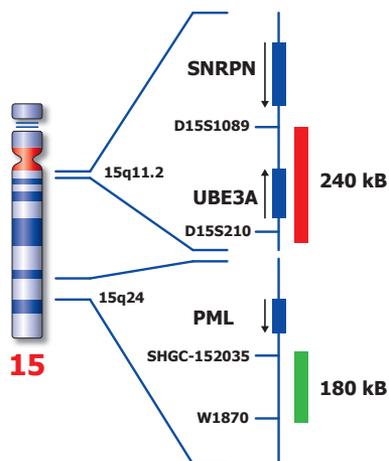
Angelman UBE3A (15q11) / PML (15q24)



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Not to scale

Kreatech™ Angelman UBE3A (15q11) / PML (15q24) FISH probe

Introduction: Angelman syndrome (AS) is characterized by four cardinal features: 1) severe developmental delay or mental retardation; 2) severe speech impairment; 3) gait ataxia and/or tremulousness of the limbs; and 4) a unique behavior with an inappropriate happy demeanor that includes frequent laughing, smiling, and excitability. In addition, microcephaly and seizures are common. All cases of clinically diagnosed AS are caused by the loss of the maternally inherited imprinted gene UBE3A (previously known as AS, ANCR, HPVE6A) in the chromosome 15q11-q13 region. Patients with AS with large chromosome deletions appear to be more severely affected than those patients with uniparental disomy. Both syndromes may also result from uniparental disomy which is not detectable by FISH analysis.

Intended use: The **Angelman UBE3A** region probe is optimized to detect copy numbers of the UBE3A gene region at 15q11. The **PML** (promyelocytic leukemia) gene specific FISH probe at 15q24 is included as 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 **Angelman UBE3A** specific FISH probe is direct-labeled with PlatinumBright™550.
Control region 2 (green): The **PML** FISH probe is direct-labeled with PlatinumBright™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 **Angelman UBE3A (15q11) / PML (15q24)** FISH probe is designed as a dual-color assay to detect deletions at 15q11. Deletions involving the UBE3A gene region will show one red signal and two green signals at the PML (15q24) control region (1R2G). Two single color red and green signals will identify the normal chromosomes 15 (2R2G)

	Normal Signal Pattern	Del(15q11) UBE3A
Expected Signals	2R2G	1R2G

References: Kishino T. et al, 1997, Nat Genet 15; 70-3
 Burger et al, 2002, Am J Med Genet. 111(3); 233-237
 Christian SL et al, 1998, Genome Res. 8(2); 146-157

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 formaldehyde 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.