Kreatech™ FISH probes

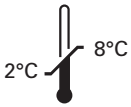
Product Information Sheet

KBI-45008

RCAN1 (21q22), SE X, SE Y

IVD

CE 2797



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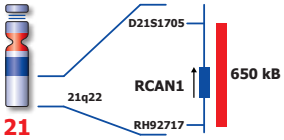
Published August 2020

PI-KBI-45008_D2.0



Not to scale

KBI-45008

Kreatech™ RCAN1 (21q22), SE X, SE Y FISH probe

Intended purpose:

The intended purpose of the device is to aid in the diagnosis of a trisomy 21, and/or copy number aberrations of the sex chromosomes X and Y using uncultured amniotic cells as a sample. The probes are intended to be used in a semi-quantitative fluorescence in situ hybridization (FISH) assay to determine the following:
The **RCAN1 (21q22)** specific FISH probe is optimized to detect copy numbers of chromosome 21 at 21q22 on uncultured amniotic cells.
The **SE X (DXZ1)** (Satellite Enumeration) FISH probe is optimized to detect copy numbers of chromosome X at Xp11-Xq11 on uncultured amniotic cells.
The **SE Y (DYZ3)** (Satellite Enumeration) FISH probe) is optimized to detect copy numbers of chromosome Y at Yp11-Yq11 on uncultured amniotic cells.

Warnings and Limitations:

This product is not intended for use on any type of cells/tissue other than uncultured amniotic cells. This test should never be used as a standalone test, but always in conjunction with other results/follow-up testing.
This FISH assay will not detect the presence of structural chromosome abnormalities that can also result in birth defects.

Introduction:

Trisomy 21 is one of the most common chromosomal abnormalities in live born children and causes Down syndrome, a particular combination of phenotypic features that includes mental retardation and characteristic facies. Molecular analysis has revealed that the 21q22.1-q22.3 region appears to contain the gene(s) responsible for the congenital heart. Chromosomal abnormalities involving the X and Y chromosome (sex chromosomes) are slightly less common than autosomal abnormalities and are usually much less severe in their effects. The high frequency of people with sex chromosome aberrations is partly due to the fact that they are rarely lethal conditions. **Turner** syndrome occurs when females inherit only one X chromosome; their genotype is X0. **Metafemales** or triple-X females, inherit three X chromosomes; their genotype is XXX or more rarely XXXX or XXXXX. **Klinefelter** syndrome males inherit one or more extra X chromosomes; their genotype is XXY or more rarely XXXY, XXXXY, or XY/XXY mosaic. **XYY** syndrome males inherit an extra Y chromosome; their genotype is XYY.

Critical region 1 (red): The **RCAN1 (21q22)** FISH probe is direct-labeled with PlatinumBright™550.
Critical region 2 (green): The **SE X** FISH probe is direct-labeled with PlatinumBright™495..
Critical region 3 (blue): The **SE Y** FISH probe is direct-labeled with PlatinumBright™415.

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 **RCAN1 (21q22), SE X, SE Y** FISH probe is designed as a triple-color assay to detect aneuploidies of chromosome 21, X, and Y in a single hybridization assay. In females two single color red (R) and green (G) signals will identify the normal chromosomes 21 and X (2R2G). In males two single colors red (R) and one green (G) and one blue (B) signals will identify the normal chromosomes 21, X, and Y (2R1G1B). Deviations in the number of sex chromosomes will be detected by more or less signals of the X- or Y- chromosome than normal (as shown below); see also Indications for use.

Expected Signals	Female	Male
Normal	2R2G	2R1G1B
Trisomy 21	3R2G	3R1G1B
XO	2R1G	-
XXX	2R3-5G	-
XXY		2R2G1B 2R3-4G1B 2R1G1B/2R2G1B in mosaics

References:

Korenberg J. et al, 1994, Proc. Nat. Acad. Sci. 91; 4997-5001
Spathas D et al, 1994, Prenat Diagn. 14(11); 1049-1054
Tepperberg et al, 2001, Prenat Diagn 21(4); 293-301

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 6919181or 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.