Kreatech™ FISH probes

Product Information Sheet

KBI-40006

RB1 (13q14)/RCAN1 (21q22), SE X (DXZ1) / SE Y (DYZ3) / SE 18 (D18Z1)

IVD

CE 2797

DANGER

FORMAMIDE

2°C 8°C

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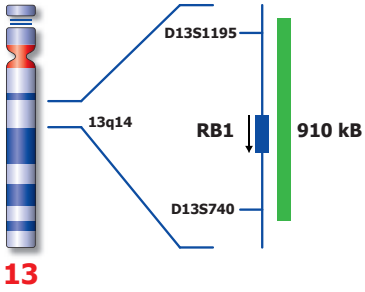

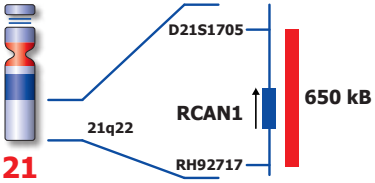

Not to scale

KBI-40006

Kreatech™ RB1 (13q14)/RCAN1 (21q22), SE X (DXZ1) / SE Y (DYZ3) / SE 18 (D18Z1) FISH probes

Intended purpose:

The intended purpose of the device is to aid in the diagnosis of a trisomy 21, trisomy 13, and/or trisomy 18 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 of numbers of chromosome 21 at 21q22 on uncultured amniotic cells.
The **RB1 (13q14) specific** FISH probe is optimized to detect copy of numbers of chromosome 13 at 13q14 on uncultured amniotic cells.
The **SE 18 (D18Z1)** (Satellite Enumeration) FISH probe is optimized to detect copy of numbers of chromosome 18 at 18p11-18q11 on uncultured amniotic cells.
The **SE X (DXZ1)** FISH probe is optimized to detect copy of numbers of chromosome X at Xp11-Xq11 on uncultured amniotic cells.
The **SE Y (DYZ3)** FISH probe is optimized to detect copy of 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.

Indications for use:

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 disease observed in Down syndrome.
Trisomy 13, also called Patau syndrome, is a chromosomal condition that is associated with severe mental retardation and certain physical abnormalities. The critical region has been reported to include 13q14-13q32 with variable expression, gene interactions, or interchromosomal effects. **Trisomy 18** causing Edwards syndrome is the second most common autosomal trisomy after trisomy 21. The disorder/condition is characterized by severe psychomotor and growth retardation, microcephaly, microphthalmia, malformed ears, micrognathia or retrognathia, microstomia, distinctively clenched fingers, and other congenital malformations. 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.

Vial 1
Critical region 1 (red): The **21q22 specific** FISH probe is direct-labeled with Platinum*Bright*™550.
Critical region 2 (green): The **13q14 specific** FISH probe is direct-labeled with Platinum*Bright*™495.
Vial 2
Critical region 3 (blue): The **SE 18** FISH probe is direct-labeled with Platinum*Bright*™415.
Critical region 4 (green): The **SE X** FISH probe is direct-labeled with Platinum*Bright*™495.
Critical region 5 (red): The **SE Y** FISH probe is direct-labeled with Platinum*Bright*™550.

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 **RB1 (13q14)/RCAN1 (21q22)** FISH probe is designed as a dual-color assay to detect gains of chromosome 21 and 13. Trisomy 21 will be detected by three red signals at the 21q22 region and two green signals for chromosome 13 (3R2G). Trisomy 13 will be detected by 3 green signals at the 13q14 region and two red signals for chromosome 21 (2R3G). Two single color red and green signals will identify the normal chromosomes 13 and 21 (2R2G).
The **SE X (DXZ1) / SE Y (DYZ3) / SE 18 (D18Z1) specific** FISH probe is designed as a triple color assay to detect gains or losses of chromosome X, Y and/or 18. Two single green signals will identify the normal X chromosomes in females (2G). One green and one red signal will identify the normal X and Y chromosomes in males (1R1G). Trisomy 18 will be detected by three blue signals for chromosome 18 (3B). Two single blue signals will identify the normal chromosome 18 (2B). 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 Using	Normal Signal Pattern		Trisomy 21	Trisomy 13	Trisomy 18	
13/21	2R2G		3R2G	2R3G		
	Female	Male			Female	Male
X/Y + 18	2G2B	1R1G2B			2G3B	1R1G3B

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

Reagent Storage and Handling:

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

Safety data sheet: 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.

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.