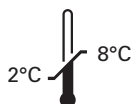


Kreatech™ FISH probes

Product Information Sheet

KBI-40103

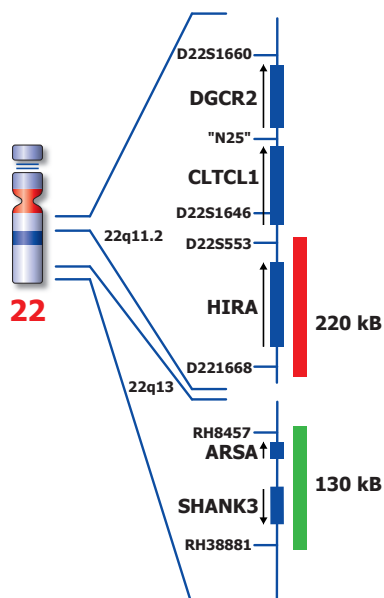
DiGeorge HIRA (22q11) / 22q13 (SHANK3)



Kreatech Biotechnology B.V.
Vierweg 20
1032 LG Amsterdam
The Netherlands
www.LeicaBiosystems.com

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Not to scale

Kreatech™ DiGeorge HIRA (22q11) / 22q13 (SHANK3) FISH probe

Introduction:

Deletions of chromosome **22q11** is the most frequent known interstitial deletion in man. Over 90% of patients with DiGeorge syndrome (DGS) or velocardiofacial syndrome (VCFS) have a microdeletion at 22q11.2. There is a wide spectrum of clinical variability from the more severe DGS to VCFS, conotruncal anomaly, abnormal facies and isolated congenital heart disease. The clinical variability is not related to the extent of the deletion since nearly all patients have the same 2 Mb deletion. The collective acronym CATCH22 (Cardiac abnormality/Abnormal facies, T-cell deficit due to thymic hypoplasia, Cleft palate, Hypocalcemia resulting from 22q11 deletions) has been proposed for all these differing presentations and covers all with common genetic etiology. The **22q13** deletion syndrome (or Phelan-McDermid syndrome) is characterized by moderate to profound mental retardation, delay/absence of expressive speech, hypotonia, normal to accelerated growth, and mild dysmorphic features. A terminal deletion including the SHANK3 gene region has been identified for this syndrome.

Intended use:

The **DiGeorge HIRA** region probe is optimized to detect copy numbers of the HIRA (previously known as TUPLE1, DGCR1) gene region at 22q11. The **22q13 FISH** probe is optimized to detect copy numbers of the SHANK3 gene region at 22q13.

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): Critical region 2 (green):

The **DiGeorge HIRA (22q11)** specific FISH probe is direct-labeled with PlatinumBright™550.
The **22q13 (SHANK3)** specific 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 **DiGeorge HIRA (22q11) / 22q13 (SHANK3)** FISH probe is designed as a dual-color assay to detect deletions at 22q11 and 22q13. Deletions involving the HIRA region will show one red signal and two green signals (1R2G). Deletions involving the 22q13 (SHANK3) region will show one green signal and two red signals (2R1G). Two single color red and green signals will identify the normal chromosomes 22 (2R2G).

	Normal Signal Pattern	Del(22q11) HIRA	Del(22q13) SHANK3
Expected Signals	2R2G	1R2G	2R1G

References:

DiGeorge, A. M. 1968, Birth Defects Orig. Art. Ser. IV(1): 116-121
Mattei, M.-G et al, 1994, Genomics 23: 717-718
Roberts, C. 1997, Hum. Molec. Genet. 6: 237-245
Scambler, P. 2000, Human Molecular Genetics, 9: 2421-2426
Wilson, et al, 2003, J Med Genet 40: 575-584
Luciani, et al, 2003, J Med Genet 40: 690-696

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.