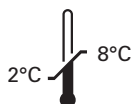


Kreatech™ FISH probes

Product Information Sheet

KBI-40104

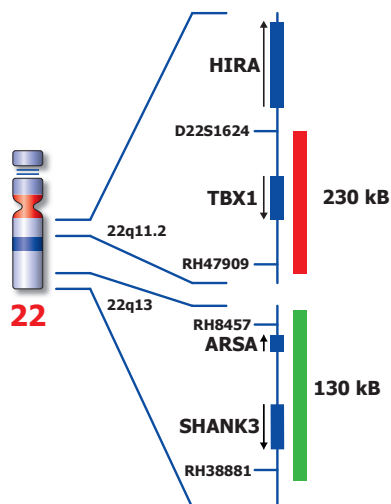
DiGeorge TBX1 (22q11) / 22q13 (SHANK3)



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

PI-KBI-40104_D1.1

Published March 2015



Not to scale

Kreatech™ DiGeorge TBX1 (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. Tbx1, a member of the T-box transcription factor family, is required for normal development of the pharyngeal arch arteries in a gene dosage-dependent manner and is sufficient to generate at least one important component of the DiGeorge syndrome phenotype in mice. The DiGeorge TBX1 probe covers this gene and adjacent regions. 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 **TBX1** region probe is optimized to detect copy numbers of the TBX1 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):** The **DiGeorge TBX1** specific FISH probe is direct-labeled with PlatinumBright™550.
Critical region 2 (green): 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 TBX1 (22q11) / 22q13 (SHANK3)** FISH probe is designed as a dual-color assay to detect deletions at 22q11 and 22q13. Deletions involving the TBX1 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) TBX1 | Del(22q13) SHANK3 |
|------------------|-----------------------|-----------------|-------------------|
| Expected Signals | 2R2G | 1R2G | 2R1G |
- References:** DiGeorge, A, 1968, Birth Defects Orig. Art. Ser. IV(1); 116-121
 Lindsay et al, 2001, Nature 410; 97-101
 Liao et al, 2004, Human Molecular Genetics 13; 1577-1585
 Luciani, et al, 2003, J Med Genet 40; 690-696
 Wilson et al, 2003, J Med Genet 40; 575-584
- 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.