Cat Nr/REF: KBI-40104

English

For professional use only

Poseidon™ Repeat Free™ DiGeorgeT-box1 (22q11) & ST 22qter Control 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.

Intended use: The **DiGeorge TBX1** region probe is optimized to detect copy numbers of the TBX1 gene region at 22q11.2.
The Sub-Telomeric (ST) 22qter DNA probe is included as control probe.

The probe is recommended to be used in combination with a Poseidon FISH Kit providing necessary reagents to perform FISH (KBI-60002, KBI-60003 or KBI-60001) for optimal results.

Critical region 1 (red): The **DiGeorge TBX1** specific DNA probe is direct-labeled with Platinum*Bright*550.

Critical region 2 (green): The **ST 22qter** control DNA probe is direct-labeled with Platinum*Bright*495.

Reagent: Poseidon 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 Poseidon FISH protocol.

Poseidon Repeat Free probes do not contain Cot-1 DNA. Hybridization efficiency is therefore increased and background, due to unspecific binding, is highly reduced.

Interpretation: The **DiGeorge Tuple (22q11)** probe is designed as a dual-color assay to detect deletions at 22q11. Deletions involving the DiGeorge region will show one red signal and two green signals at the 22qter control region (1R2G). Two single color red (R) and green (G) signals will identify the normal chromosomes 22 (2R2G)

	Normal Signal Pattern	Del 22(q11) TBX1
Expected Signals	2R2G	1R2G

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

POSEIDON™
REPEAT-FREE™ FISH PROBES

Application Manual

KBI-40104
MD DiGeorge T-box1 (22q11) / 22qter

IVD
for EU only

CE

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2°C 8°C long term storage

KREATECH DIAGNOSTICS

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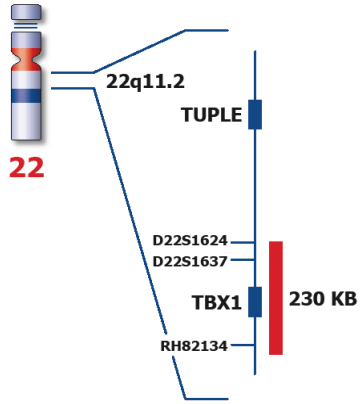

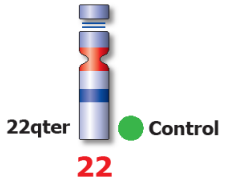

Not to scale

Application manual