

## Poseidon™ Repeat Free™ Chromosome 13 (13q14) & 21 (21q22) Specific probes

**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 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.

**Intended use:** The **chromosome 21 specific** region probe is optimized to detect copy numbers of chromosome 21 at 21q22.1 on uncultured amniotic cells. The **chromosome 13 specific** region probes is optimized to detect copy numbers of Chromosome 13 at 13q14.2 on uncultured amniotic cells.

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 **21q22 specific** DNA probe is direct-labeled with PlatinumBright550.

**Critical region 2 (green):** The **13q14 specific** DNA probe is direct-labeled with PlatinumBright495.

**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 **Chromosome 13/21 specific** probe is designed as a dual-color assay to detect gains of chromosome 21 and 13. Trisomy 21 will be detected by three red signal 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 (R) and green (G) signals will identify the normal chromosomes 13 and 21 (2R2G).

	Normal Signal Pattern	Trisomy 21	Trisomy 13
Expected Signals	2R2G	3R2G	2R3G

**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

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## Application Manual

KBI-40003

PN 13 (13q14) / 21 (21q22)



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