

Poseidon™ Repeat Free™ Chromosome 13/21, XY/18 specific DNA 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.

**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.

**XXY syndrome** males inherit an extra Y chromosome; their genotype is XYY.

**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 **chromosome 18 specific** Satellite probe (D18Z1) is optimized to detect copy numbers of Chromosome 18 at 18p11-18q11 on uncultured amniotic cells.

The **chromosome X specific** Satellite probe (DXZ1) is optimized to detect copy numbers of Chromosome X at Xp11-Xq11 on uncultured amniotic cells.

The **chromosome Y specific** Satellite probe (DYZ3) is optimized to detect copy numbers of Chromosome Y at Yp11-Yq11 on uncultured amniotic cells.

These probes are recommended to be used in combination with the Poseidon FISH & Digestion Kit (KBI-60003) providing necessary reagents to perform FISH for optimal results.

- Vial 1**  
**Critical region 1 (red):** The 21q specific DNA probe is direct-labeled with PlatinumBright550.  
**Critical region 2 (green):** The 13q14 specific DNA probe is direct-labeled with PlatinumBright495.
- Vial 2**  
**Critical region 3 (blue):** The 18 SE DNA probe is direct-labeled with PlatinumBright415.  
**Critical region 4 (green):** The X SE DNA probe is direct-labeled with PlatinumBright495.  
**Critical region 5 (red):** The Y SE DNA probe is direct-labeled with PlatinumBright550.

More information on the backside

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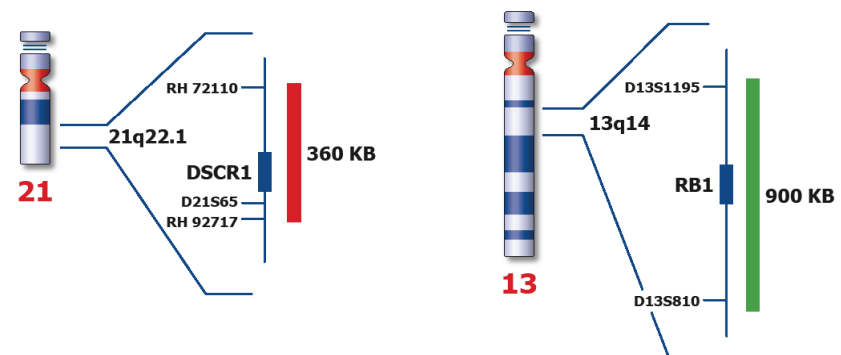
# Application Manual

KBI-40005  
 PrenatScreen (13/ 21, X/ Y/18)



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Application manual