



## **New! Marker for the diagnosis of Turner Syndrome**

November 2010

Turner syndrome is most commonly caused by the absence of one X chromosome in a female and diagnosed as a 45,X0 karyotype.

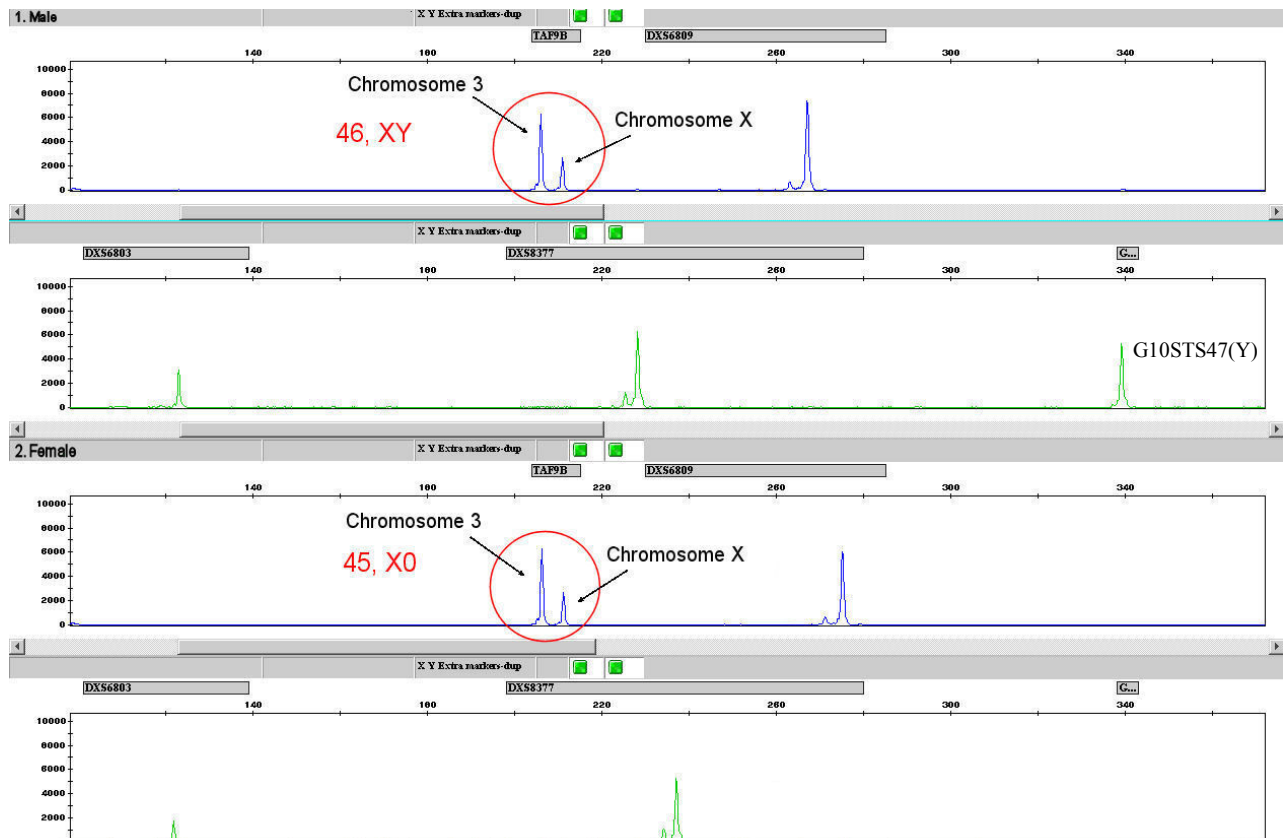
A new marker called TAF9B has been introduced into the ChromoQuant X,Y Extra marker kit.

**TAF9B** is a non polymorphic and stable marker with sequences found in the TAF9B gene on chromosome X, as well as in its pseudogene on chromosome 3.

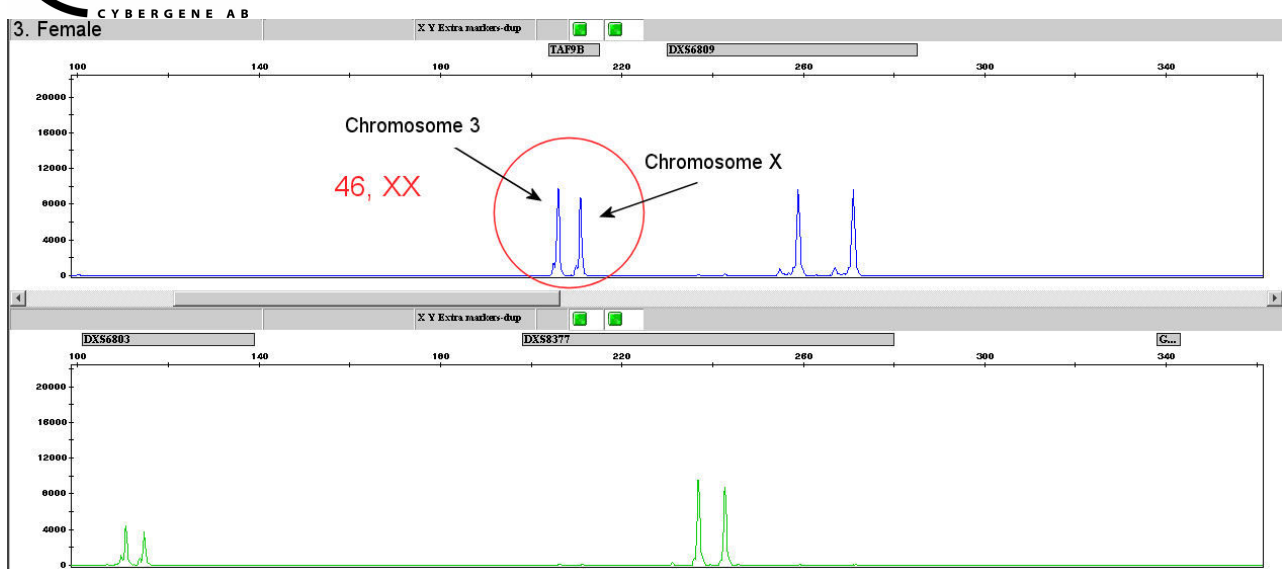
The specific peak for TAF9B on chromosome 3 is 205 bp, and represents 2 alleles.

The specific peak for TAF9B on chromosome X is 210 bp, and represents 2 alleles in a normal female (46,XX karyotype). In Turner syndrome (45,X0 karyotype in a female), this marker will represent only 1 allele. The chromosome 3 specific peak can therefore be used as a reference peak when determining the number of X chromosomes present.

TAF9B should be used in combination with the other sex chromosome markers present in the kit, like the chromosome Y-specific marker G10 STS47, when diagnosing Turner syndrome. In a female Turner sample (45,X0 karyotype) or normal male (46,XY karyotype), the peaks from the TAF9B marker give a ratio  $\geq 1.8$ . In a normal female (46, XX karyotype) the peaks from the TAF9B marker give ratios between 0.8 and 1.4. TAF9B can also be used together with the other sex chromosome markers when determining sex chromosome aneuploidies like XXY or XYY.



**Sample 1: Normal male sample;** For TAF9B: 2 alleles of chromosome 3, 1 allele of chromosome X; ratio 1:2. Single alleles for the other X markers. Presence of chromosome Y-specific marker G10 STS47. **Sample 2: Turner female sample;** For TAF9B: 2 alleles of chromosome 3, 1 allele of chromosome X; ratio 1:2. 1 allele for the other X markers. Absence of chromosome Y-specific marker G10 STS47.



**Sample 3: Normal female sample;** For TAF9B: 2 alleles of chromosome 3 and X; ratio 1:1. 2 alleles for the other X markers. Absence of chromosome Y-specific marker G10 STS47.