The Combined Power of Karyotyping and aCGH

WiCell Cytogenetics Laboratory offers BOTH assays for in-depth characterization of human pluripotent stem cell lines.

Chromosome 2

The 24.15 Mb deletion on chromosome 2 of the cell line is visible by both g-banded karyotyping and aCGH (gp168,490,000-192,639,990)

Resolution — karyotyping detects genomic abnormalities >5-10 Mb
Specificity — aCGH confirms and provides accurate breakpoints

Chromosome 4

The 1.4 Mb gain on chromosome 4 of the same cell line is visible by aCGH only (gp94,430,000-95,830,000)

Resolution — aCGH allows identification of small genomic abnormalities (>50kb)

Chromosome 12

The 3 copies of chromosome 12, a recurrent abnormality in iPSCs and hESCs, are visible in 2 of 20 cells by g-banded karyotyping, but not by aCGH

Sensitivity — Unlike g-banded karyotyping, aCGH does not consistently detect genomic abnormalities present at low level mosaicism (<20%) within the culture

G-banded karyotyping is the gold standard for assessing cell line cytogenetic stability based on a maximum resolution of 5-10 Mb, while array Comparative Genomic Hybridization (aCGH) is a powerful technique for identification of small genomic abnormalities greater than 50 kb. WiCell Cytogenetics Laboratory offers g-banded karyotyping and aCGH testing including:

- Custom analysis
- Experienced CNV profiling of hESC lines
- Well-characterized reference DNA

This case was an hES cell line submitted for cytogenetic analysis with the resulting karyotype: 46,XX,del(2)(p24q32)[7]/47,XX,del(2)(p24q32),+12[1]/47,XX,+12[1]/46,XX[11]