WiCell Cytogenetics Laboratory

Guidelines for cytogenetic testing of cell lines

Assay	What it Detects	What it Doesn't Detect	TAT[i]	When to Use
G-Banded Karyotyping	• Microscopic genomic abnormalities (>5-10 Mb) - Inversions - Duplications/deletions - Balanced and unbalanced translocations - Aneuploidies • >10% mosaicism (for example: cultures where >1 of 10 cells are trisomy 12)	• Submicroscopic genomic abnormalities (<5 Mb)	7-10 days	• As a baseline genomic screen - At derivation of cell lines - At the start of experimental protocols - To assess and monitor genomic stability (for example: every 5-10 passages of cell culture) - At conclusion of experiments (prior to publication) - For cell line banking • When publication-quality karyotypes are needed
Spectral Karyotyping (SKY)	• Microscopic genomic abnormalities (>5-10 Mb) - Balanced and unbalanced translocations - Aneuploidies	• Submicroscopic abnormalities (<5 Mb) • Inversions • Duplications/deletions	10-15 days	• As an adjunct to g-banded karyotyping - To define complex rearrangements - To identify marker chromosomes • When publication-quality spectral karyotypes are needed
Microarray by SNP and/or CGH	• Microscopic genomic abnormalities (>5-10 Mb) • Genomic gains and losses (>50 Kb) - Copy number variants - Duplications/deletions - Unbalanced translocations - Aneuploidies • >~10% mosaicism (for example: cultures where >1 of 10 cells are trisomy 12 • Loss of Heterozygosity (LOH) / Absence of Heterozygosity (AOH) - detected by SNP	• Balanced translocations • Inversions • <~10% culture mosaicism (for example: cultures where 1 of 10 cells is trisomy 12) • Chromosomal position of genomic gains • Loss of Heterozygosity (LOH) / Absence of Heterozygosity (AOH) - not detected by CGH alone	14-21 days	• As a baseline genomic screen - To detect submicroscopic (<5 Mb) abnormalities - To identify amplified or deleted genes of interest • In conjunction with g-banded karyotyping - To define unbalanced translocation breakpoints • For research of genomic copy number change - To identify structural variation within populations or disease cohorts - To develop a cell line copy number variant profile
Fluorescence In Situ Hybridization (FISH)	• Genomic sequence of interest - Duplications or deletions >20 Kb - >2% mosaicism (for example: cultures where >2 of 100 cells are trisomy 12) - Chromosomal location of genomic gains - Chromosome fusions (breakaparts)	• Changes in regions other than the probe-specific sequence	10-15 days	• To confirm findings and refine breakpoints detected by g-banded karyotyping • To confirm findings and localize genomic gains detected by aCGH analysis • As a screen for microdeletions/duplications of known targets
Fast FISH	• Greater than 50% of recurrent trisomies in both human ES and iPS cell lines.[ii] - Trisomies of human chromosomes X, 8, 12 and 17.	• Aneuploidies of chromosomes other than X, 8, 12 and 17 • Structural chromosome abnormalities	1-2 days	• As a rapid screen for large numbers of clones • As a cost-effective tool for monitoring aneuploidy
Short Tandem Repeat Analysis (STR)	• STR polymorphisms for 15 loci plus amelogenin (Promega® PowerPlex® 16) • Probability of matching identity to an existing STR profile	• STR polymorphisms in areas other than those represented in Promega® PowerPlex® 16	10-15 days	• To monitor identity of a cell line • To confirm relationship of iPS cells to their parent line • To establish an STR profile of a newly derived or reprogrammed cell line • To rule out culture cross-contamination

[i] Turn-around times provided are based on provision of sufficient mitotically active hES or iPS cultures grown in matrigel/TeSR or MEF/hES media conditions.
[ii] Based on n=1220 hES/iPS cell cultures karyotyped by WiCell Cytogenetics Laboratory.

Your Lab Partner

09052013