Table A5.2. Common recurrent genetic changes in hPSCs, some of their phenotypic consequences and suitable methods for their detection

Recurrent Genetic Change	Acquired as...	Some of the Reported Phenotypes of Variant Cells	Suitable Methods for Testing	Comments
Chromosome 1q gain	Gain of the whole chromosome (trisomy 1), whole q arm or an interstitial duplication.		Karyotyping (e.g., G-banding or low pass sequencing)*; qPCR or ddPCR; SNParrays or aCGH arrays; eSNP-karyotyping; FISH	*In some instances, the gain of 1q is acquired as an interstitial duplication below the resolution of karyotyping
Chromosome 12 gain	Gain of the whole chromosome 12 (trisomy 12), isochromosome 12p or a gain of the whole or parts of the p arm.	In undifferentiated state, trisomy 12 cells exhibit increased proliferation and teratocarcinoma formation (Ben-David et al., 2014); Variants with a 12p gain also show reduced ability for differentiation (Keller et al., bioRxiv)	Karyotyping (e.g., G-banding or low pass sequencing)*; qPCR or ddPCR; SNParrays or aCGH arrays; eSNP-karyotyping; FISH	*In some instances, the gain of 12p is acquired as an interstitial duplication below the resolution of karyotyping.
Chromosome 17q gain	Gain of the whole chromosome 17 or a gain of the whole or parts of the q arm	Variants with a chromosome 17q gain exhibit selective growth advantage, supercompetitive phenotype in high cell density cultures (Price et al., 2021) and altered differentiation patterns (Lee et al., 2015)	Karyotyping (e.g. G-banding or low pass sequencing); qPCR or ddPCR; SNParrays or aCGH arrays; eSNP-karyotyping; FISH	

Chromosome 18q loss	An interstitial deletion of q arm		Karyotyping (e.g., G-banding or low pass sequencing)*; qPCR or ddPCR; SNParrays or aCGH arrays; FISH	*Deletion may be below the resolution of karyotyping
Chromosome 20q gain	Gain of the whole chromosome 20 (trisomy 20), gain of isochromosome 20q or an interstitial duplication	Selective growth advantage in undifferentiated state (Avery et al., 2013; Nguyen et al., 2014) reduced genetic stability (Zhang et al., 2019) and altered differentiation patterns (Markouli et al., 2019; Werbowetski-Ogilvie et al., 2009)	Karyotyping (e.g., G-banding or low pass sequencing)*; qPCR or ddPCR; SNParrays or aCGH arrays; FISH	Karyotyping can detect trisomy 20 and isochromosome 20q, but interstitial duplications of 20q are often below the resolution of karyotyping
Single nucleotide variants in TP53	Typically acquired in DNA-binding domain of TP53 as dominant negative mutations	Selective growth advantage in undifferentiated state; also selected for during some differentiation protocols (Merkle et al., 2017)	Next-generation sequencing (DNA or RNA) Sanger sequencing	