

Supplementary Table

Individual results of genotyping and FISH experiments at 17q21.31 in cell lines from 7 individuals of European descent, 7 HapMap subjects of European descent, and in 3 17q21 deletion patients and their parents

Cell line ID	Genotype	FISH status*	Sample Description
7 individuals of European descent			
GM17207	H2/H2	Heterozygous	Human Variation Panel, Coriell
GM17219	H2/H2	Inversion	Human Variation Panel, Coriell
GM17251	H2/H2	Heterozygous	Human Variation Panel, Coriell
GM17225	H2/H2	Heterozygous	Human Variation Panel, Coriell
GM17258	H2/H2	Heterozygous	Human Variation Panel, Coriell
GM17249	H2/H2	Inversion	Human Variation Panel, Coriell
GM17202	H2/H2	Inversion	Human Variation Panel, Coriell
7 HapMap subjects of European descent			
GM06994	H1H2	Heterozygous	CEPH1340, paternal grandfather
GM07000	H1/H1	Direct	CEPH1340, paternal grandmother
GM07019	H1/H1	Direct	CEPH1340, mother
GM06985	H1H2	Heterozygous	CEPH1341, maternal grandmother
GM06991	H1/H1	Direct	CEPH1341, mother
GM10851	H1/H1	Direct	CEPH1344, father
GM10861	H1/H1	Direct	CEPH1362, mother
Patients + parents			
Nijm31874	H1/H1	Direct	Parent proband 31873
Nijm31875	H1/H2	Heterozygous	Parent proband 31873
Nijm31873	H1/ -	Direct / deletion	Proband with 17q21 deletion
Nijm31918	H2/H2	Heterozygous	Parent proband 31920
Nijm31919	H1/H1	Direct	Parent proband 31920
Nijm31920	H1/ -	Direct / deletion	Proband with 17q21 deletion
Nijm31944	H1/H2	Heterozygous	Parent proband 31928
Nijm31927	H1/H1	Direct	Parent proband 31928
Nijm31928	H1/ -	Direct / deletion	Proband with 17q21 deletion

*FISH status reported as “Direct” when the orientation corresponds with “wild type”.