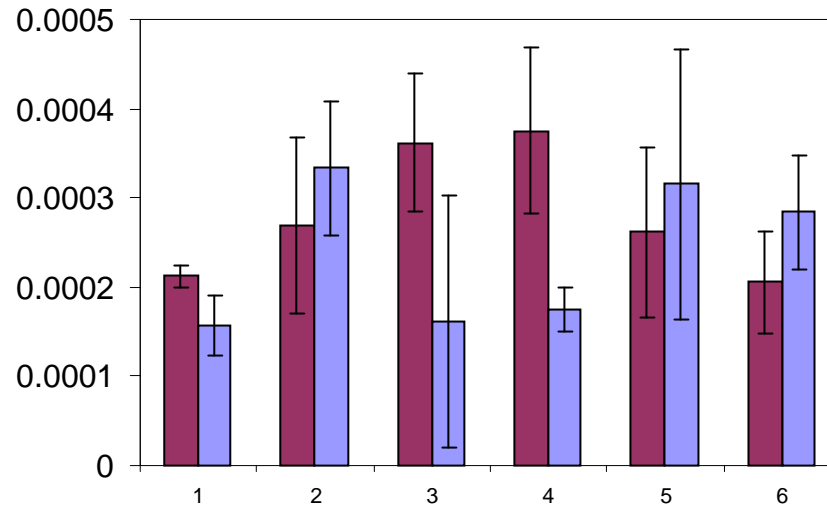| Pair #	Patient Cell Lines			Relative Cell Lines		
	Repository #	**Sex**	**Age**	**Repository #**	**Relation**	**Age**
1	GM14162	M	28	GM14163	Mother	55
2	GM14171	M	38	GM14173	Father	68
3	GM14182	M	22	GM14183	Father	55
4	GM14268	F	19	GM14267	Mother	49
5	GM14291	F	11	GM14292	Mother	39
6	GM14297	F	32	GM14295	Mother	62

Supplementary Table S1.
Coriell Cell Repository Cell lines
Cell line repository number (#), sex and age of the patients from which the lymphoblastoid cell lines were derived. The Pair number (#) corresponds to the pairing shown on the graphs (Fig. 3 and Fig. S1). Full details of the cell lines and the patients from which they are derived is available at http://ccr.coriell.org

Name	Sequence
mIRD1ex1F	CCAGAGGCGACTGGATCTAC
mIRD1ex3R	ACGAAGACGCTCTCATTGTG
mIRD1ex6F	TACAGCACTTCGATGCCCAACCAC
mIRD1ex7/8R	GATCCCACTTCTCTGACTTGTCATG
mHPRTF	AGCTTGCTGGTGAAAAGGAC
mHPRTR	TCAACTTGCGCTCATCTTAG
hCYLN2F	AAATCCCTGTCGGATCAGAG
hCYLN2R	TTTGCAGAACCGGAATTGCC
hGTF2IF	CTGGAAATCAGCTCCATGAG
hGTF2IR	GATCTGAGAGCTTCCATCAG
hIRD1ex21F	TGGAGAGGATTCTTGCTGTG
hIRD1ex23R	CCTCACCGTATTTCTCGTTG
hIRD1ex25F	GGCTGGAGAAGATCCTGAAG
hIRD1ex27R	ATGCTGCTGTCTTTGGCTGG
hGAPDHF	CCATGTTCGTCATGGGTGTG
hGAPDHR	CATCCACAGTCTTCTGGGTG
hIRD1LZdel	TTAAGCTTACCATGGACTTCCTCAGGTTCTGCCG
GURF	TGCGGGCTAATCCCGTATGTAAATG
GURF MUT	TGCGGGCT**TGA**CCCGTATGTAAATG
GURR	TTTGTTCTGTAATCCCAGTAAGATTG
GURR MUT	TTTGTTCTGT**TGA**CCCAGTAAGATTG
GUR MID MUT	TGCGGGCTAATCCCGTATGTAAATGACAGCCAATGGAGGGTGGTGTTGCGCGGGGC TGGG**TCA**AGGGCCGGGGCGAATGGCTGGCAATCTTACTGGGATTACAGAACAAA
GUR LUC F	GAGTCTCGAGGCTCCGAGGTTG
GUR LUC R	GCGAGCGCGAAGCCGAGCCCG

Supplementary Table S2 Names and sequence of the oligonucleotides synthesised for all experiments. Bold type indicates the position and sequence of the mutant residues.

Supplementary Figure S1
A second analysis of GTF2IRD1 expression in lymphoblastoid cell lines.
Quantitative RT-PCR analysis of *GTF2IRD1* expression in lymphoblastoid cell lines derived from WBS patients and their relatives (see Table S1) using the primers Ex25F – Ex27R (see Table S2). Magenta bars represent the relative samples and blue bars represent patient samples. Error bars show standard deviations.