

A

		Gender	Age
C1	27574-B	M	3,5
C2	95-3985-S	F	7
C3	95-3999-B	M	8
C4	GIA-001-S	F	6,8

		Gender	Age	Mutation
PT1	132-01P	F	9	c.4606C>T/ Nonsense
PT2	173-04P	M	6,9	c.6407_6408 insA/ Frameshift
PT3	285-05P	M	0	c.2449_2450insG/ Frameshift

C

Pyrosequencing of mRNA from CdLs LCLs:

Treatment	DMSO		Cycloheximide		
	Allele	WT	MUTANT	WT	MUTANT
PT1 (c.4606C.T)		C: 62%	T: 38%	C: 52%	T: 48%
PT3 (c.2449 2450insG)		C: 76%	T: 24%	C: 54%	T: 46%

NIPBL-AS1 and NIPBL mRNA levels in control and patient LCLs:

B

