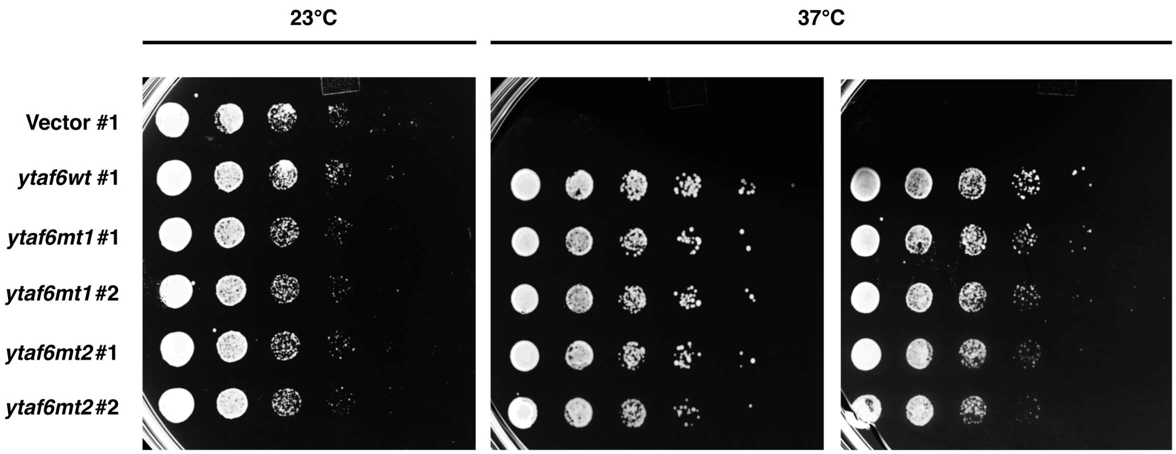
Supplemental Data



Supplemental figure 1 – Yeast spot assay of the two *TAF6* mutations identified in CdLS-4 and the Saudi patients. Vector, empty vector used as negative control; *ytaf6wt*, vector with yeast wild type *TAF6* used as positive control; *ytaf6mt1*, vector with yeast *TAF6* mutant corresponds to $hTAF6N^{R46C}$; *ytaf6mt2*, vector with yeast *TAF6* mutant corresponds to $hTAF6N^{I71T}$. For groups of *ytaf6mt1* and *ytaf6mt2*, two parallel assays (#1 and #2) are performed.

Supplemental Table 1. Potential modifier mutation identified in the family of WDSTS-1/2.

Patient ID	**Zygosity**	**Clinical diagnosis**	**Genomic coordinates**	**nt change**[A]	**aa change**[B]	**Gene**	**Conserved**	**Predicated**	**Frequencies**		
									ESP[C]	TGP[D]	ARIC[E]
BAB4135 (WDSTS-1)	Homozygous	WDSTS	Chr20: 58455447	c.T2852C	p.I951T	*SYCP2*	+	Damaging	0.23%	0.05%	0.5%
BAB4136 (WDSTS-2)	Heterozygous										

[A] Nucleotide change;
[B] Amino acid change;
[C] Exome Variant Server;
[D] 1000 Genomes project;
[E] Internal control database of ~4000 exomes at Baylor College of Medicine.

Supplemental Table 2. The gene targeted on the CdLS aCGH with their names chromosomal and genomic locations (hg18) listed accordingly.

Name of the Gene	Chromosome	Location (hg18)
PLK3	chr1	45038623-45044254
SMC6	chr2	17708560-17798577
NCAPH	chr2	96365211-96405001
SGOL2	chr2	201099110-201157063
SHOX2	chr3	159296494-159306630
SGOL1	chr3	20177089-20202687
STAG1	chr3	137538689-137953935
SMC4	chr3	161600124-161635435
NLGN1	chr3	174598938-175483810
NAALADL2	chr3	176059805-177006122
SOX2	chr3	182912416-182914917
CHRD	chr3	185580555-185590311
NCAPG	chr4	17421623-17455585
PDS5A	chr4	39500878-39655971
NIPBL	chr5	36912618-37101678
PTTG1	chr5	159781443-159788324
STAG3	chr7	99613474-99649946

NCAPG2	chr7	158116764-158190281
ESCO2	chr8	27687977-27718343
RAD21	chr8	117927355-117956286
SMC5	chr9	72063698-72159609
SMC2	chr9	105896362-105943521
WAPAL	chr10	88184993-88271521
SMC3	chr10	112317439-112354382
CDCA5	chr11	64601503-64608191
ATM	chr11	107598769-107745036
NCAPD3	chr11	133527547-133599636
ESPL1	chr12	51948350-51973694
CBX5 (HP1A)	chr12	52910997-52960182
TIMELESS	chr12	55096425-55129467
PDS5B	chr13	32058592-32250157
REC8	chr14	23711074-23719303
GSC	chr14	94304313-94306252
TIPIN	chr15	64416062-64436108
PLK1	chr16	23597702-23609189
CTCF	chr16	66153965-66230589

AURKB	chr17	8048774-8054608
ESCO1	chr18	17363260-17434691
KIAA0892	chr19	19292614-19330564
KIF3B	chr20	30329128-30386472
CTCFL	chr20	55505630-55533560
PTTG1IP	chr21	45093941-45118169
SMC1B	chr22	44118609-44188164
NCAPH2	chr22	49293511-49305057
SMC1A	chrX	53417795-53466343
STAG2	chrX	122922156-123064186

Supplemental Table 3. The primers for Sanger confirmation and segregation analysis.

No.	Families	Forward primers		Reverse primers	
		Name	Sequence (5'-3')	Name	Sequence (5'-3')
1	BAB3616	NIPBL-26F	TTTTCTGGCTTTCTTAAAATCTG	NIPBL-26F	GCTCACAAGCATCCAGAATC
2	BAB3617	NIPBL-10F1	GAGCTTTCAAAGAGTGAAATGAAAC	NIPBL-10R1	TCGTTCTGATTTTAACCGAGG
3	BAB3621	NIPBL-2F	TTGTGTGTTGCAGTGTTTGG	NIPBL-2R	TTAAAGGGCAGTTTCAGTTGC
4	BAB3623	SMC1A-5F	ACCTCTTGGCTCTGAGGGAC	SMC1A-5R	ATAAACAGCACGGCCTCTTG
5	BAB3967	NIPBL-10F2	CATTGATCTTCATCAGGCAGGA	NIPBL-10R2	GGCCTTGATTTTGTTGTTTCTG
6	BAB4008	NIPBL-7F	TGGAATCTGGCAGAGTGTTAAG	NIPBL-7R	TATGTGCAGGAGCCATGTTG
7	BAB4037 (CdLS-4)	TAF6-2F	TCTCCACTGTCCTCCCTTTG	TAF6-2R	AGACCTGGCTGAAGGATGG
8	BAB4135/4136 (WDSTS-1/2)	SMC1A-2F	TTGATGAGAATGCCCAACTTC	SMC1A-2R	TGTTGTCCAAGCTGGTTTCA
		SYCP2-30F	GACGAAAATCATGAGCCAAGGA	SYCP2-30R	TGATCTGGTTTTCCAAGTGACTTTC
9	BAB4141	NIPBL-3F	TTTGTTAGGAAGAGGAGGAATG	NIPBL-3R	TTTCTGAAATAAAACCAGGAATACG
10	BAB4695	NIPBL-20F	GAAAACATTTTCATTCTAAATGGC	NIPBL-20R	TGACGGTTCAATATAATGGTGG
11	BAB4709	NIPBL-9F	CATCTTTCGTAAATAAGTAGCTTGG	NIPBL-9R	TTGTTCTTGCTGCGACAAAC
12	BAB4797	NIPBL_46F	ACCCACACCAAACTACTGCC	NIPBL_46R	AAGGATGTATAAACGGGTAAGTAGAG
13	BAB4938	HDAC8_4F	TCACACCTCAACCTATTCCTCC	HDAC8_4R	CACACACGTCTCTCTGTAAATGTC
14	BAB4964 (CdLS-3)	KMT2A_3F	GCATCTGGTTTTTCTGCATCTG	KMT2A_3R	TGAGTCAGGGAATGAGAAGGAA
15	BAB5452 (CdLS-2)	SMC1A-22F	TGTCTCGGACACTTTCCAGCTTT	SMC1A-22R	CTGGGGGAAGGGCTAGAGATTAGA
		ABCA4-42F	CCCCAGGGTAAGTGTGTGTGTG	ABCA4-42R	GTGCCTGCCCATGTCAATATGT
16	BAB5593 (CdLS-1)	SMC3-23F	TCAGTGGTGGTTAGATGCTTAGAAA	SMC3-23R	TGCTTCTGCCTATTTGGACATT
17	BAB6475	NIPBL-44F	GAAGCTTTTTCAAGCTGTTGAATGG	NIPBL-44R	GCATGCAAACAAAACTGGGTTTC
18	Saudi	TAF6-3F	GCCCAGATTCAGGAGGAGAC	TAF6-3R	CATAGAAGTAAAGCTCCCGGC

Supplemental Table 4. The primers for splicing site mutation analysis.

Families	Forward primers		Reverse primers	
	Name	Sequence (5'-3')	Name	Sequence (5'-3')
BAB5452 (CdLS-2)	5452- SMC1A-F	GGTAGCCACAGATTCAAAACAAGCA	5452- SMC1A-R	AGTGAGTGGTTCACAGAGAATTTCCA
BAB5593 (CdLS-1)	5593-SMC3-F	ACTGGGAACTGATTTGCTTTCTCAA	5593-SMC3-R	GCCGATTTGTCATCTTTTCCAGTTC