**Supplementary Materials:** Supporting data consists of supplementary method, three figures and one table.
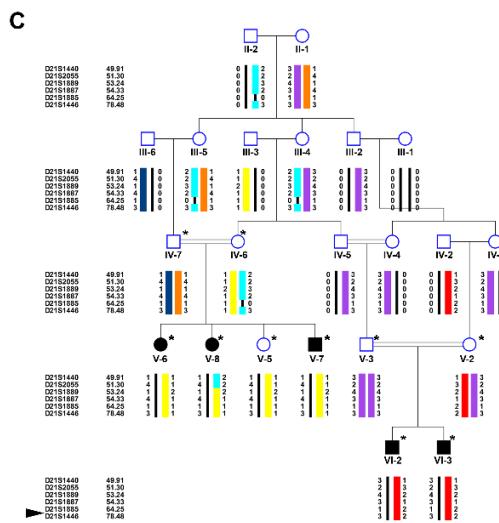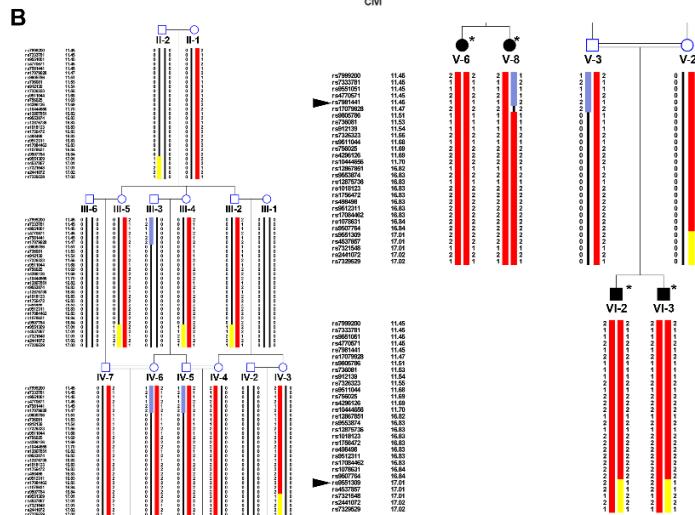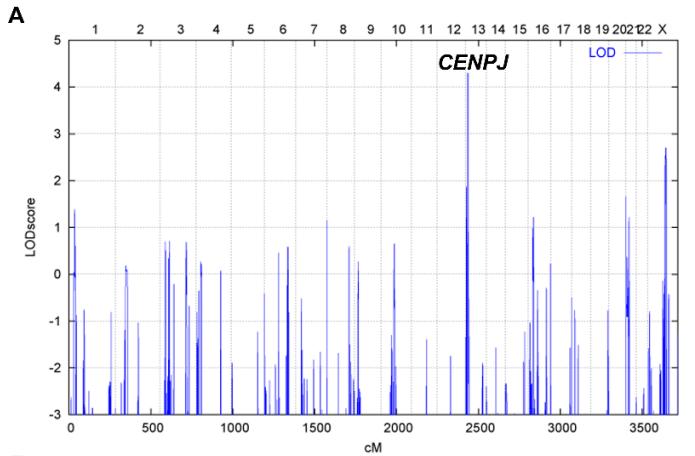
**Supplementary methods**

**Linkage analysis**

Genome-wide linkage analysis was performed using the graphical user interface ALOHOMORA[1]. Relationship errors were identified by the program Graphical Relationship Representation [2]. The program PedCheck was applied to find Mendelian errors [3], and data for SNPs with such errors were removed from the data set. Non-Mendelian errors were identified by using the program MERLIN [4], and unlikely genotypes for related samples were deleted. Linkage analysis was performed assuming autosomal recessive inheritance, full penetrance, consanguinity and a disease allele frequency of 0.0001. Multipoint LOD scores were calculated using ALLEGRO [5]. Haplotypes were reconstructed with ALLEGRO and presented graphically with HaploPainter [6].
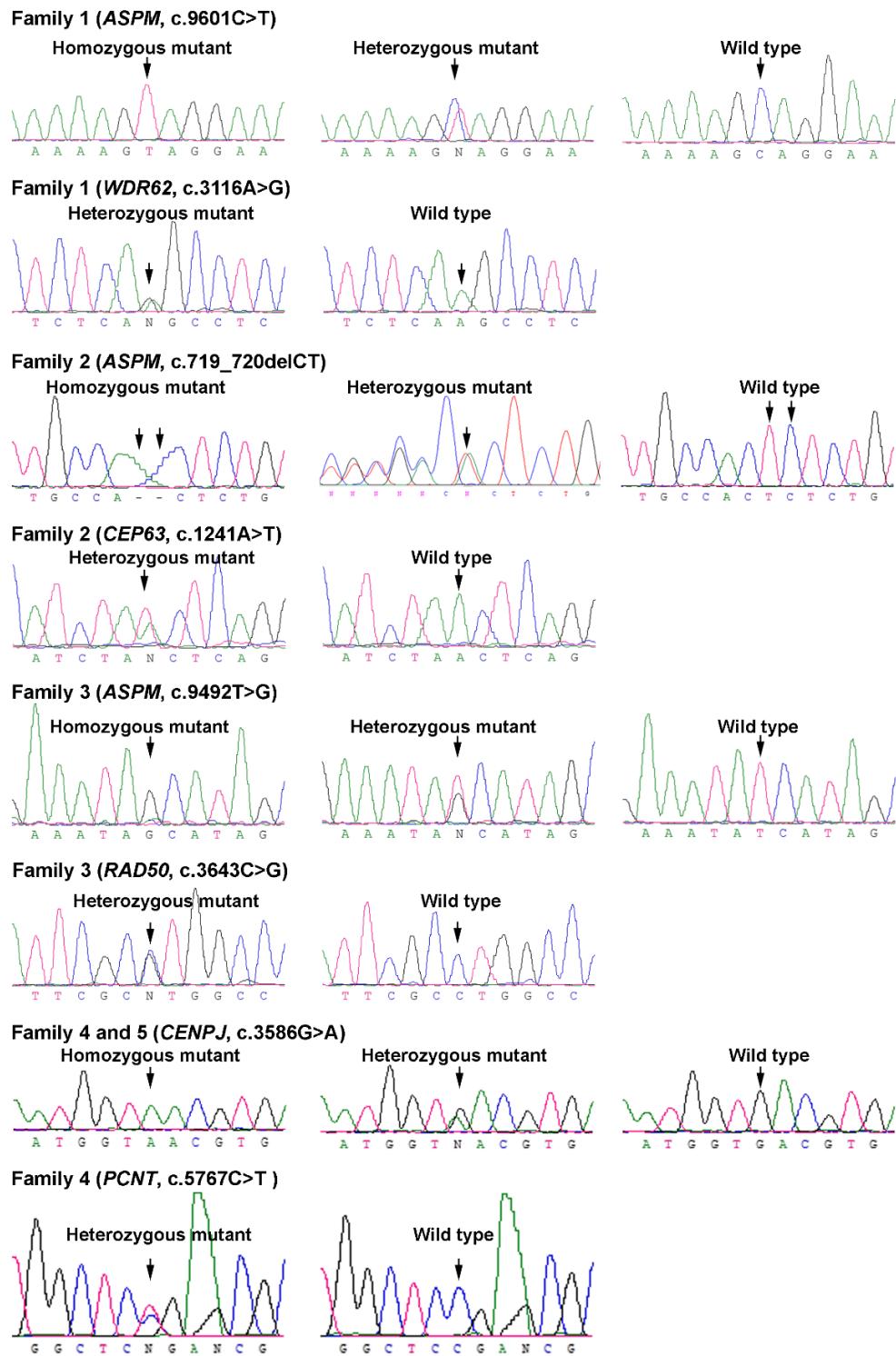
**Supplementary References**

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6.  Thiele, H.; Nurnberg, P. HaploPainter: a tool for drawing pedigrees with complex haplotypes. *Bioinformatics* **2005**, *21*, 1730-1732, doi:10.1093/bioinformatics/bth488.

**Figure S1.** Linkage and haplotype analysis of family 4. **(A)** Genome-wide LOD plot using markers equally spaced by 100 kb. The maximum LOD score of 4.3 was reached on chr 13. The homozygous region covers the *CENPJ* locus. **(B)** Haplotypes generated to show the homozygous region of approximately 2.63 Mb located at chromosome 13 harboring *CENPJ*. The haplotype

drawing is splitted into two panels. Left panel indicates the family members belonging to second, third and fourth generations whereas individuals belonging to fifth and sixth generations are shown in the right panel. Arrowheads show the recombinant markers, rs7981441 (24,075,006 bp) and rs9551309 (26,712,326 bp). Physical positions refer to human genome build GRCh38.p12. **(C)** Haplotypes constructed by genotyping microsatellite markers surrounding *PCNT*. Asterisks denote the affected members used for genotyping. The arrowhead pointing between D21S1885 (chr21:44,759,989-44,960,367 bp) and D21S1446 (chr21:47,937,583-48,129,895 bp) indicates the position of *PCNT* (47,743,976-47,865,682 bp). Physical positions refer to human genome build GRCh37.p13.

**Family 1 (*ASPM*, c.9601C>T)**

Homozygous mutant

A A A A G T A G G A A

Heterozygous mutant

A A A A G N A G G A A

Wild type

A A A A G C A G G A A

**Family 1 (*WDR62*, c.3116A>G)**

Heterozygous mutant

T C T C A N G C C T C

Wild type

T C T C A A G C C T C

**Family 2 (*ASPM*, c.719_720delCT)**

Homozygous mutant

T G C C A - - C T C T G

Heterozygous mutant

Wild type

T G C C A C T C T C T G

**Family 2 (*CEP63*, c.1241A>T)**

Heterozygous mutant

A T C T A N C T C A G

Wild type

A T C T A A C T C A G

**Family 3 (*ASPM*, c.9492T>G)**

Homozygous mutant

A A A T A G C A T A G

Heterozygous mutant

A A A T A N C A T A G

Wild type

A A A T A T C A T A G

**Family 3 (*RAD50*, c.3643C>G)**

Heterozygous mutant

T T C G C N T G G C C

Wild type

T T C G C C T G G C C

**Family 4 and 5 (*CENPJ*, c.3586G>A)**

Homozygous mutant

A T G G T A A C G T G

Heterozygous mutant

A T G G T N A C G T G

Wild type

A T G G T G A C G T G

**Family 4 (*PCNT*, c.5767C>T )**

Heterozygous mutant

G G C T C N G A N C G

Wild type

G G C T C C G A N C G

**Figure S2. Sanger traces of the identified mutations are shown.** For each family, chromatograms of the homozygous disease-causing variations are shown on top, and below are the traces of heterozygous modifying variations. Traces of *ASPM*;c.719_720delCT heterozygous mutant are shown in reverse orientation.

**A**

## WDR62, Ser1106

```
Homo sapiens              VEASEAEDH-----FFNPRLSISTQFLSSLQKASRFTHTFPPRATQCLVKSPEVKLMDRG
Struthio camelus australis LKPPEAGEE-EKTLFLNPRLSISARFLSRCQKNSRWVLNAAYPSVRPEKGLPGACWARKP
Bos taurus                LKAPETEDD-----FFNPRLSISAQFLSRLQKTSRFPHAFPSRLPQHLVKSSEVRLTDLR
Mus musculus              IKISETEDY-----FFNPRLSISRLQKTS----RCPPRLPLHLMKSPEAQPVGQG
Monodelphis domestica     VEPPEMDADSEKSFFLNSRLSMSTQFLSRFQKNSRFAHTFPPTLPLQLVKPLEAVVLRRE
Podarcis muralis          LKAPEEEEE-EAALFQNPRLSISTRFLSSCQKNSRFAAPFLPRLRQHSQAAAADQLAEES
Danio rerio               LKPTEDHGN---DIFLHPRLSISARFLSRCQKNSSFSRWYPSLFGLTSITLLKYFLSIRK
                          :    :       * :.***:*::* *    * *
```

**B**

## WDR62, Ser1106

```
Macaca mulatta            LGDVEASEAEEYFFNPRLSISTQFLSSLQKASRFTHTLPPRATQHLVKSPEVKPMDQVGS
Cercocebus atys           LGDVEASEAEEYFFNPRLSISTQFLSSLQKASRFTHTLPPRATQHLVKSPEVKPMDQVGS
Homo sapiens              --DVEASEAEDHFFNPRLSISTQFLSSLQKASRFTHTFPPRATQCLVKSPEVKLMDRGGS
Pan troglodytes           LGDVEASEAEDYFFNPRLSISTQFLSSLQKASRFTHTFPPRATQCLVKSPEVKLMDRGGS
Gorilla gorilla           LGDTEASEAEDYFFNPRLSISTQFLSSLQKASRFTHTFPPRATQCLVKSPEVKLMDRGGS
Pongo abelii              --DVEASEAEDYFFNPRLSISTQFLSSLQKASRFTHTFPPRATQRLVKSPEVKLMDRGGS
Callithrix jacchus        LGDTEASEAEDYYFNPRLSISTQFLSSHQKASRFTHTFPSRATQHLVKPPEVKPTDRGRS
Microcebus murinus        LRDMKVSEAEDYYN-PRLSISAQFLSSLRKTSRFPHTFPTRLPQRLVTSPEVKPTARRRS
                          * :.****::: ******:***** :*:***.**:*.* .* **..****   : *
```

**C**

## Cep63, Thr421

```
Homo sapiens              KMEISHLTQELHQRDITIASTKGSSSDMEKRLRAEMQKAEDKAVEHKEILDQLESLKLEN
Pongo abelii              KMEISHLTQELHQRDITIASTKGSSSDMEKRLRAEMQKAEDKAVEHKEILDQLESLKLEN
Mus musculus              KMEISQLTRELHQRDITIASAKCSSSDMEKQLKAEMQKAEEKAVEHKEILSQLESLKLEN
Rattus norvegicus         KTEISQLTRELHQRDITIASAKCSSSDMERQLKAEMQKAEEKAVEHKEILSQLESLRLEN
Gallus gallus             RKEILRLTQELHQRDITIASAGGSTSDLEQRLRMEIERAERKAVEHRMILVQLETLKLEN
Xenopus laevis            RKEISQLTQELHQRDIRMASSAG--IDWERKIKAERQRAEREAAEHRMSLNALENLRQEN
Danio rerio               RKEVFQLTSELHQRDITIATLTGSTSSSIERQLRAEVERAERRASELKVTQVQLETLKLEN
                          : *: :** ******* :*:      . *:::: * ::** .* * :    **.*: **
```

**D**

## hRAD50, Leu1215

```
Homo sapiens              QKVLASLIIRLALAETFCLNCGIIALDEPTTNLDRENIESLAHALVEIIK
Mus musculus              QKVLASLIIRLALAETFCLNCGILALDEPTTNLDRENIESLAHALVEIIK
Monodelphis domestica     QKVLASLIIRLALAETFCLNCGILALDEPTTNLDRENIESLAHALVEIIK
Gallus gallus             QKVLASLIIRLALAETFCLNCGILALDEPTTNLDRENIESLAHALVEIIK
Xenopus laevis            QKVLASLIIRLALAETFCLNCGILALDEPTTNLDRENIESLAHALVEIIK
Danio rerio               QKVLASLIIRLALAETFCLNCGILALDEPTTNLDRENIESLAHALVEIIK
Caenorhabditis elegans    QKMLASLLIRIALAEVFGGSCSMIALDEPTTNLDESKVEGMAIVLADIIA
Drosophila melanogaster   QRVLASLIIRLALAETFSSNCGVLALDEPTTNLDRANINSLCEALNCIVE
Arabidopsis thaliana      QKVLASLIIRLALAETFCLNCGILALDEPTTNLDGPNSESLAGALLRIME
Saccharomyces cerevisiae  QKVLASIIIRLALSETFGANCGVIALDEPTTNLDEENIESLAKSLHNIIN
                          *::***::**:**:*.* .*.::********** : :.:.  *  *:
```

**E**

## CENP-J, Asp1196

```
Homo sapiens              KVYKNGCRVILFPNGTRKEVSADGKTITVTFFNGDVKQVMP-DQRVIYYYAAAQTTHTTY
Bos taurus                KVYKNGCHVVLFPNGTRKEVSADGETVTVTFFNGDIKQVMP-DGRVVYYYAATQTAHTTF
Mus musculus              KIYKNGRRVVLFPNGTRKEVSADGKSVTVTFFNGDVKQVMP-DERVVYYYAAAQTTHTTY
Gallus gallus             KVLKNGCHLIFLPNGTWKKVGSDGKTVTITFFNGDVKQIMP-DQTVIYYYADAKTTHTTY
Danio rerio               KMLPDGGRLVVFPNGTRKELSADGQTVKVMFFNGDVKHTMP-DQRVIYYYAEAQTTHITY
Latimeria chalumnae       QVLRSGHRIIVFPNGTRKEVSSDGKTVTVTFFNGDIKQIMP-DQRVIYYYADAKTTHTTY
Xenopus tropicalis        RILRNGYHVIEFPNGTRKEVSADGKSTTVKFFNGDVKQVLG-DQRIIYYYADDQTTHTTY
Monosiga brevicollis      TILPDGTQVIEFPNGTVKKVFADGR-VHVAFFNGDTKETHA-DGRVVYFYQEAGTKHTTL
Octopus bimaculoides      KVYQNGARELIFSNGTRKEMSADGKHIALHFFNGDIKQITA-DQTVIYFYAESETLVTTY
Spizellomyces punctatus   RIYANGIKLVRYSNGTLKEVHPDDELIVIHFTNGDSKTVFA-DGRIVYWYTEQRTLHTTY
Drosophila melanogaster   ITNADGSKDIWYPNGNLKKISADGMNLRMLYFNKDIKETNIREGTVKYYYAETNTWHTSY
                          :* : :  .**. *:: .*.  : : : * * *    : : *:*     *   :
```

**Figure S3. Multiple alignments of short peptide sequences of WDR62, Cep63, hRAD50 and CENP-J from different species.** Sites of the mutant residue are colored in red. **(A)** UniProtKB IDs of WDR62 from different organisms are as follows, O43379_HUMAN, XP_009666581 (Ostrich), A0A4W2FHS7_BOBOX, Q3U3T8_MOUSE, F6UN22_MONDO, A0A670J9V7_PODMU and F6NPK9_DANRE. **(B)** UniProtKB and NCBI reference sequences IDs of WDR62 from selected primates have the following IDs, F7CAC3_MACMU, A0A2K5MJV0_CERAT, O43379_HUMAN, H2QG55_PANTR, G3QFQ3_GORGO,

H2NYJ6_PONAB, U3BAJ3_CALJA and XP_012630407. 1_MICMU. **(C)** UniProtKB IDs of Cep63 are Q96MT8_HUMAN, Q5NVN6_PONAB, Q3UPP8_MOUSE, Q4KLY0_RAT, P0CB05_CHICK, B9V5F5_XENLA and Q6PGZ0_DANRE. **(D)** UniProtKB and NCBI reference sequence IDs of hRAD50 are Q92878_HUMAN, P70388_MOUSE, XP_007473506. 1_MONDO, Q5W4T6_CHICK, A0A1L8GXM0_XENLA, F8W411_DANRE, O44199_CAEEL, Q9W252_DROME, Q9SL02_ARATH and P12753_YEAST. **(E)** UniProtKB IDs of CENP-J are as follows, Q9HC77_HUMAN, E1BL95_BOVIN, Q569L8_MOUSE, E1C0R8_CHICK, E7FCY1_DANRE, H2ZX87_LATCH, F7C9Z3_XENTR, A9V5C9_MONBE, A0A0L8G7N5_OCTBM, A0A0L0HNY0_SPIPN and Q9VI72_DROME.

**Figure S4.** Immunoblot showing the reduced amount of CENP-J in both patients (VI-2 and V-8) compared to wild type.