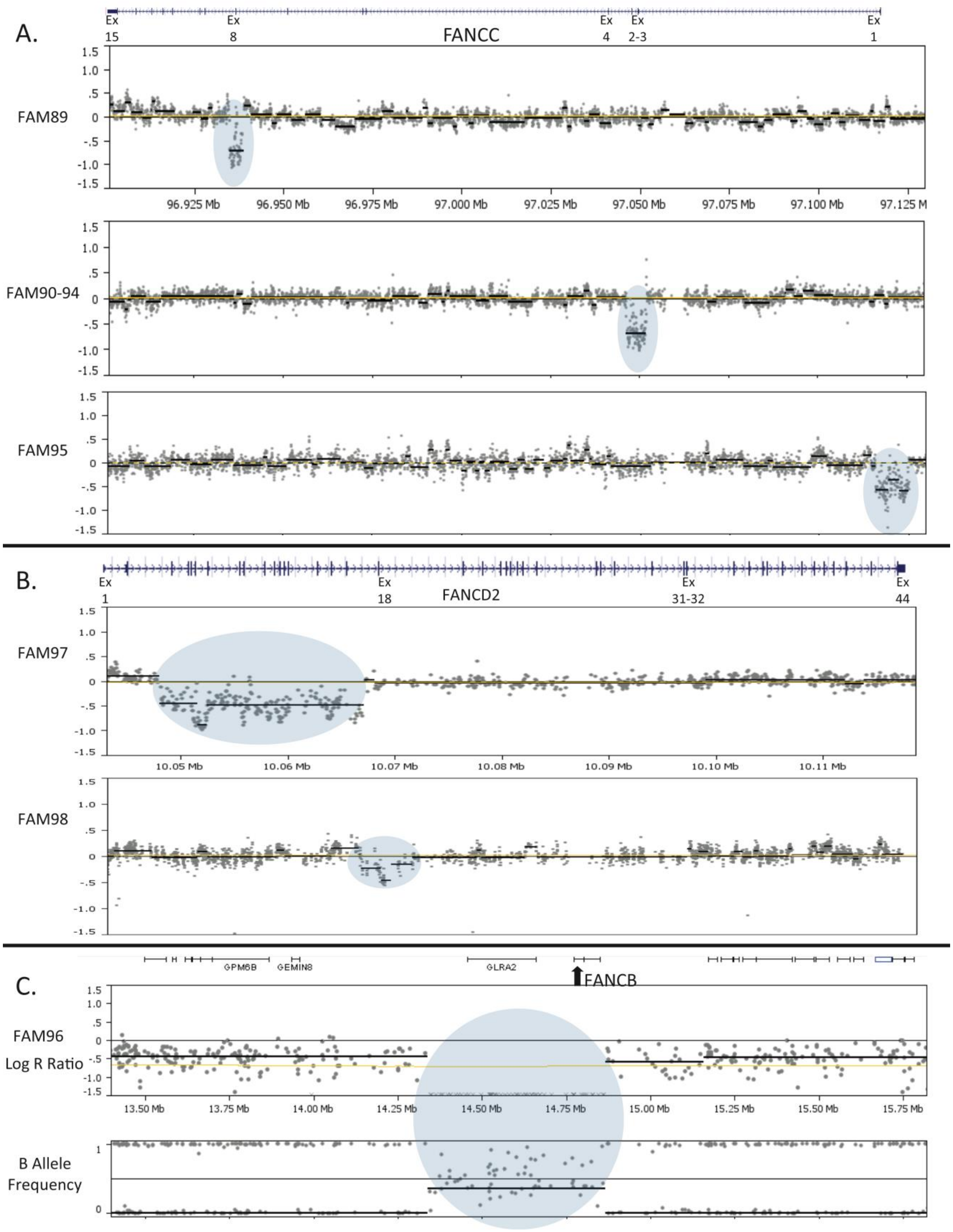
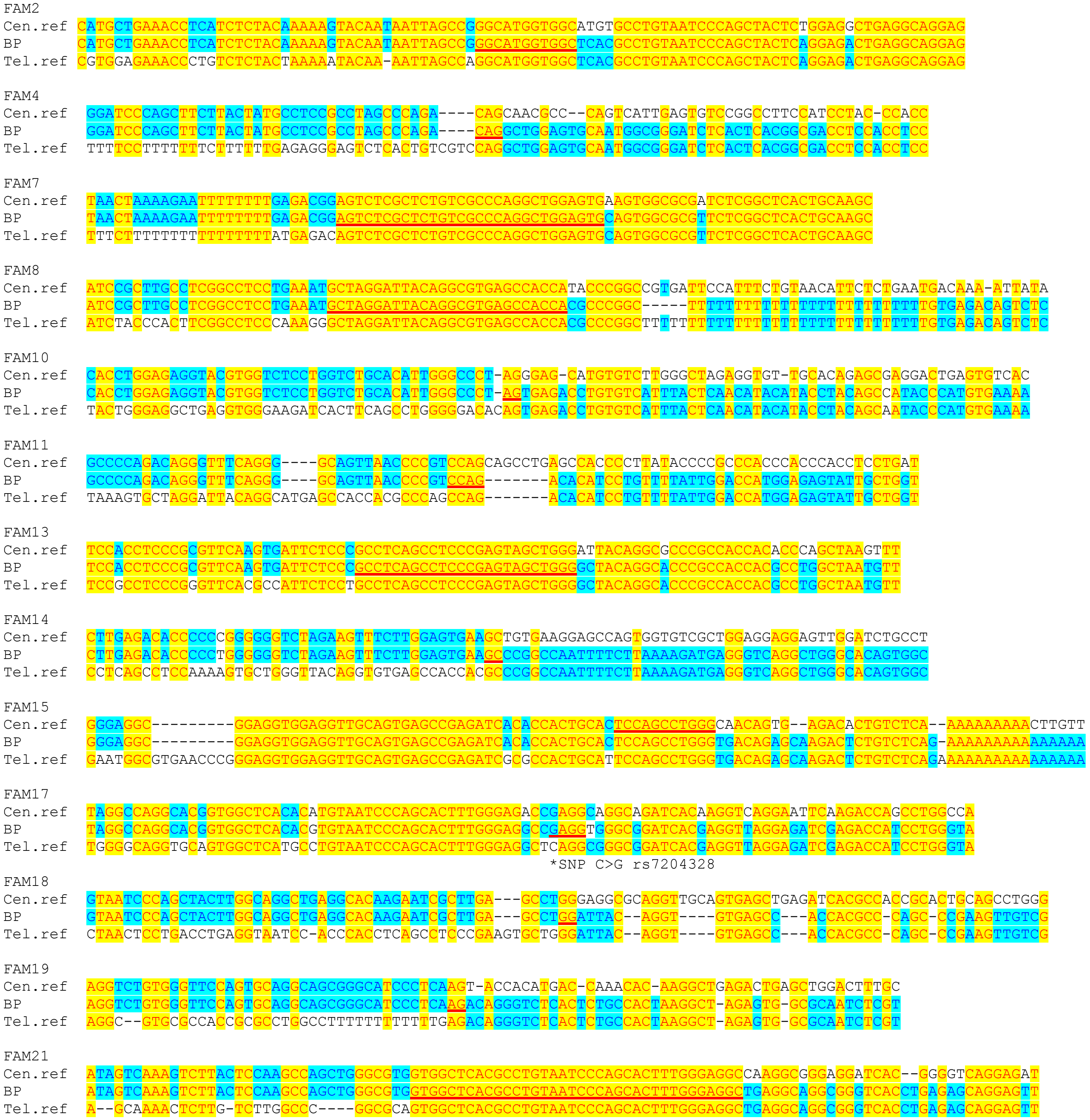
Supp. Figure S1



Supp. Figure S1. Deletions in *FANCC*, *FANCD2* and *FANCB* from aCGH and SNP array analysis. **A**. aCGH-*FANCC*. Log2 Ratio plots of comparative genomic hybridization intensities from probes in the *FANCC* region of chromosome 9. **B**. aCGH-*FANCD2*. Log2 Ratio plots of comparative genomic hybridization intensities from probes in the *FANCD2* region of chromosome 3. **C**. SNParray-*FANCB*. The top plot, Log R Ratio, shows the intensity values for SNPs in the *FANCB* region of chromosome X. There is a depressed signal (~ -.5) for all the SNPs on chromosome X because only one copy is present (compared to the reference genome). The lack of any signal from SNPs located in the deleted region is a result of no copies being present. The bottom plot is the B Allele Frequency of SNPs in the *FANCB* region of chromosome X. The group of SNPs scattered in the middle represent probes that could not bind to the deleted region. The surrounding SNPs are all homozygous, showing a B Allele Frequency of either 1 or 0, because there is only one X chromosome present. All data in **A, B, C** is displayed using Nexus 6.1 software (Biodiscovery), and the deleted regions are shaded blue.

Supp. Figure S2

A.



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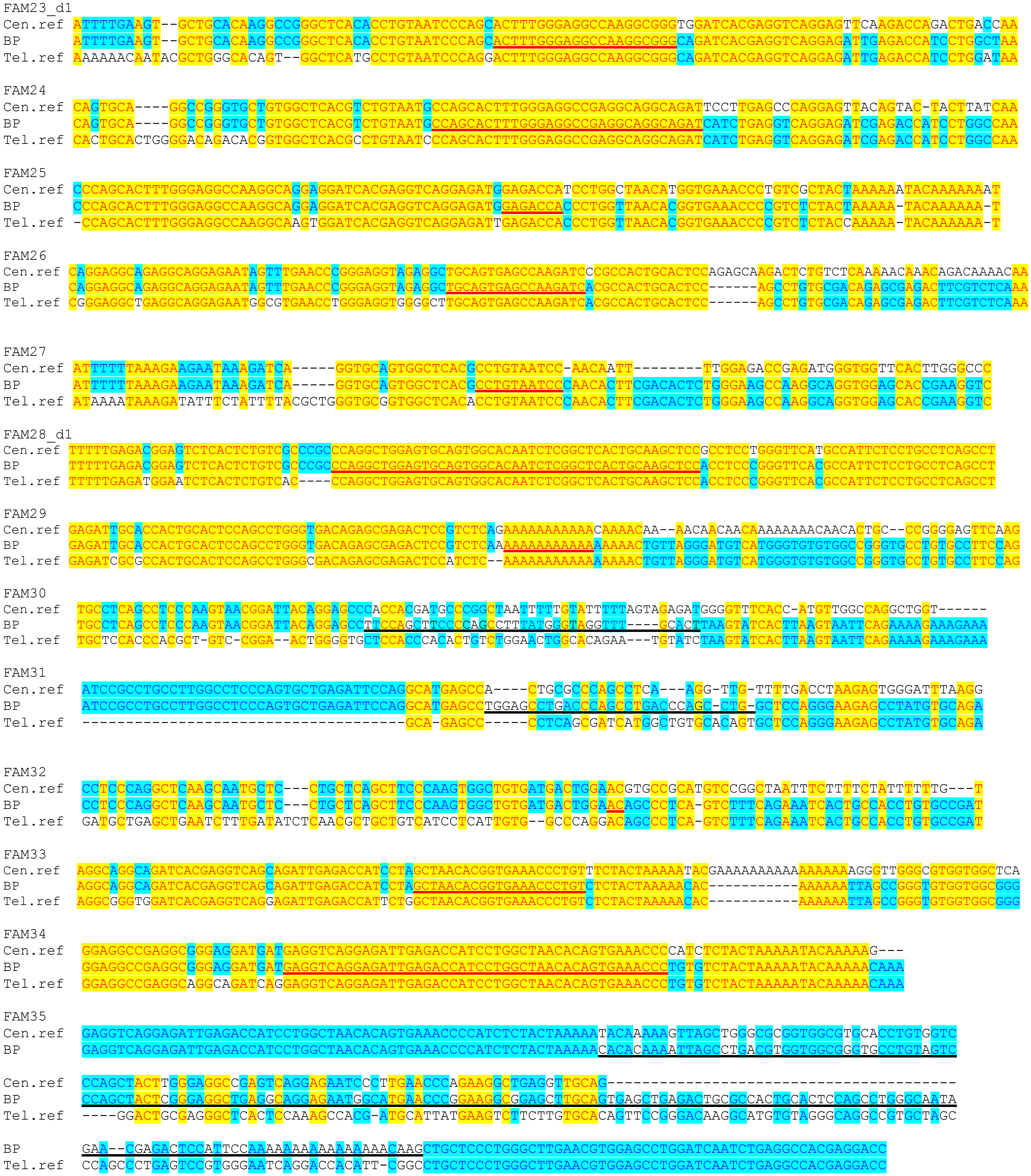

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BP        ATCAAGGGTCCAG-CCGGGCACGGTGGCTCATGCCTGTAATCCCAGCACTCTGGGAGGCCGAGGCGGGCGGATTACAAGGTCAGGAGATCGAGACCATCCTGGCTAACAC
Tel.ref   TTAAAGAAACCGGGCCGGGCACGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGCGGATTACAAGGTCAGGAGATCGAGACCATCCTGGCTAACAC

FAM67
Cen.ref   CCGTCTCTACTAAAAATACAAAAAATTAGCCGGGCGCAGTGGCGGACGCCCTGTAGTCTCAGCTACTCGGGAGGCTGAGGCAGGAGAATG
BP        CCGTCTCTACTAAAAATACAAAAAATTAGCCGGGCGCAGTGGCGGACGCC-TGTAGTCCCAGCTACTCGGGAAGCTGAGGCAGGAGAATG
Tel.ref   CAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGACGCC-TGTAGTCCCAGCTACTCGGGAAGCTGAGGCAGGAGAATG

FAM68
Cen.ref   ACTAAAAATACAAAAAAATTAGCTAGGTGCGGTGGCGGGTGTCTATAGTCCCAGCTACTCGGGAGACTGAGGCAGGAGAATAGCTTGAACCCGGTAGGCGG
BP        ACTAAAAATACAAAAAAATTAGCTAGGTGCGGTGGCGGGTGTCTATAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATGGCGTGAACCTGGGAGGTGG
Tel.ref   GCTAAAAAATACAAAAAAATTAGCTGGGCGTGGTGGCAGTCACCTGTAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATGGCGTGAACCTGGGAGGTGG

FAM71
Cen.ref   CAGATCAAAATGAGTTTTACCCAAGAACCCGCATCTTGTCATGAACGCACCAGAAA---GCATGGCCCTGGCGACGTCAGCA--TGCTGGCAGGA---TGCTTCCATC-TGTT
BP        CAGATCAAAATGAGTTTTACCCAAGAACCCGCATCTTGTCATGAACGCACCAGAACTTTGGGAGGCCGAGGCGGGTGGATCACCTGAGGTCGGGAGTTTGAGACCAGCCTGAC
Tel.ref   AAAAAAAACTGGAGGCCAGGCTCAGTGGGTCCCTCCTGTAATC------CCAGCACTTTGGGAGGCCGAGGCGGGTGGATCACCTGAGGTCGGGAGTTTGAGACCAGCCTGAC

FAM73
Cen.ref   CAGTTTTGCTCT-TGTCATCCAGGCTGGAGTGCAATGGCACAATCTCAGCTCACTACAACCTCCGCCTCCCAAGTTCAAGCGATTCTCCTGCCTCAGCCTCCACAA
BP        CAGTTTTGCTCT-TGTCATCCAGGCTGGAGTGCAATGGCACAATCTCAGCTCACGGCGACCTCCACCTCCCAGGTTCAAGCGATTCTCCTGCCTCAGCCTCCTGAG
Tel.ref   GAGGGAGTCTCACTGTCGTCCAGGCTGGAGTGCAATGGCGGGATCTCA-CTCACGGCGACCTCCACCTCCCAGGTTCAAGCGATTCTCCTGCCTCAGCCTCCTGAG

FAM74
Cen.ref   TCTTGAACTCCTGACCTCAGGTGATCTGCCCGCCTCAGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCACCGCACCTGGCCTTGGCCTGAA--CCTTATGC-
BP        TCTTGAACTCCTGACCTCAGGTGATCTGCCCGCCTCAGCCTCCCAAAGTGCTGGGATTACAGGAGTGAGCCACCGCGCTCGGCCACAAATGGAAGTCTTTATCCC
Tel.ref   TCTCAATCTCCTGACCTCT--TGATCCATCCGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGAGTGAGCCACCGCGCTCGGCCACAAATGGAAGTCTTTATCCC

FAM75, FAM76, FAM77, FAM78, FAM79, FAM80 CD5
Cen.ref   TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGACGCCTGTAGTCCCAGCTACTCGGAAGCTGAGGCAGGAGAATGGCG
BP FAM75  TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA
BP FAM76  TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA
BP FAM77  TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA
BP FAM78  TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA
BP FAM79  TAACATGGTGAA-ACCCAGTCTCTACTAAAAATACAAAAA-TTAGCCAGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA
Tel.ref   AAAATTACTAAATACAAAAAAATTACTAAAAATACAAAAAATTAGCTGGGCGTGGTGGCGGGCGCCTGTAGTCCCATCTACTTAGGAGGCTGAGGCAGGAGAATGGCA

FAM80
Cen.ref   GTCACTGTAAGCTCTGCCTCCCGGGTTCACGCCATTCTCCTGCCTCAGCCTCCTGAGTAGCTGGGACTACAGGTGCCTGCCACCACACCCGGCTAATTTTTTC
BP        GTCACTGTAAGCTCTGCCTCCCGGGTTCACGCCATTCTCCTGCCTCAGCCTCCCAAGTAGCTGGGATTACAGGCGTGTTCCTCCCGAGTAG-CTGGGATTACA
Tel.ref   -TCACCGCAACCTCCACCTCCCAGGTTCAAGCCGTTCTCTTGCCTCAGCCTCCCAAGTAGCTGGGATTACAGGCGTGTTCCTCCCGAGTAG-CTGGGATTACA

FAM81, FAM82 CD6
Cen.ref   GGATGGGCTCGATCTCCTGACCTCATGATCCACCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCACTGGGCCCGGCCCCTAACTCGCACTCTTCACAG
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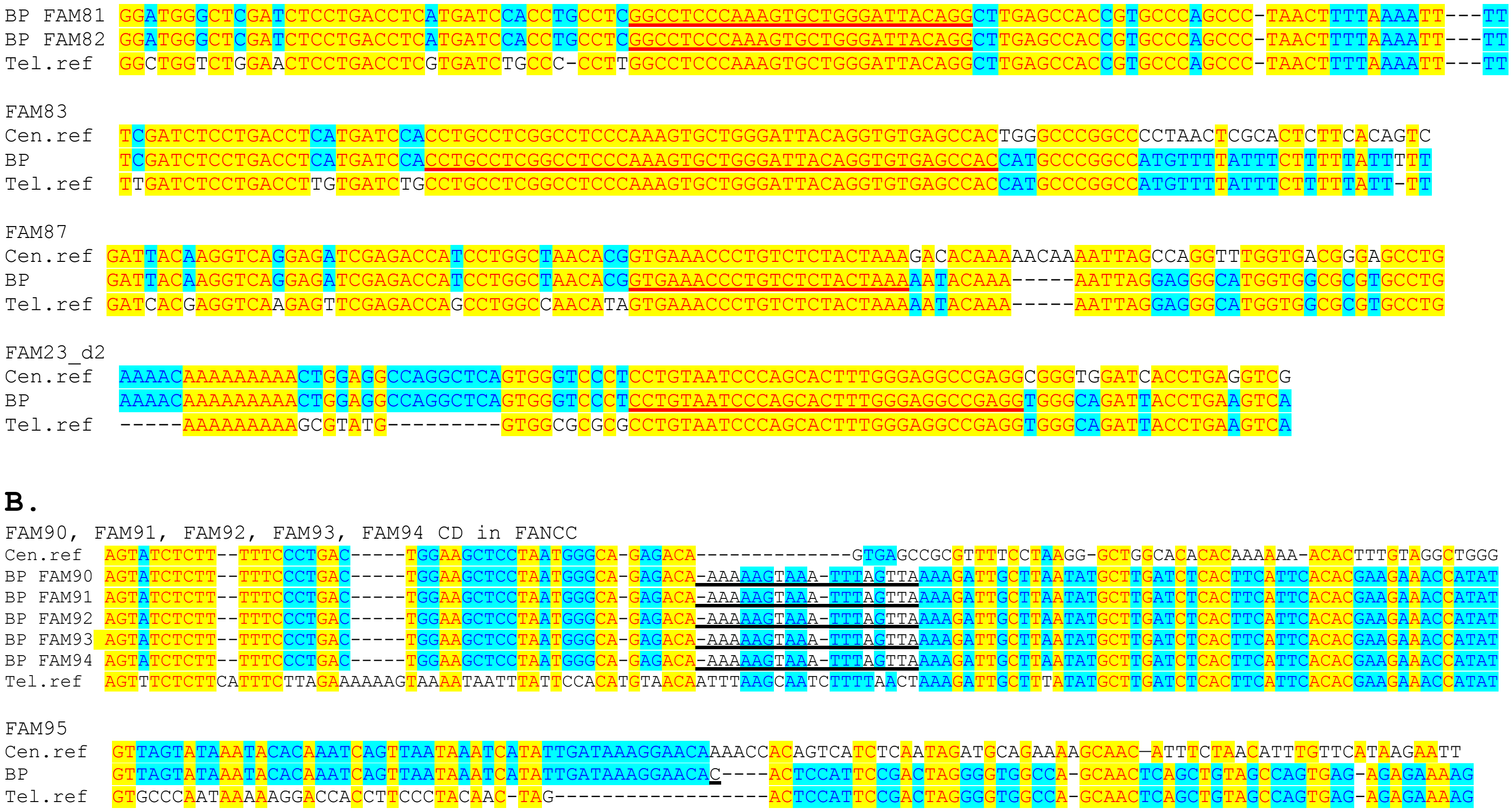

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BP FAM81 GGATGGGCTCGATCTCCTGACCTCATGATCCACCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGCTTGAGCCACCGTGCCCAGCCC-TAACTTTTAAAATT---TT
BP FAM82 GGATGGGCTCGATCTCCTGACCTCATGATCCACCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGCTTGAGCCACCGTGCCCAGCCC-TAACTTTTAAAATT---TT
Tel.ref  GGCTGGTCTGGAACTCCTGACCTCGTGATCTGCCC-CCTTGGCCTCCCAAAGTGCTGGGATTACAGGCTTGAGCCACCGTGCCCAGCCC-TAACTTTTAAAATT---TT

FAM83
Cen.ref  TCGATCTCCTGACCTCATGATCCACCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCACTGGGCCCGGCCCCTAACTCGCACTCTTCACAGTC
BP       TCGATCTCCTGACCTCATGATCCACCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCACCATGCCCGGCCATGTTTTATTTCTTTTTATTTTT
Tel.ref  TTGATCTCCTGACCTTGTGATCTGCCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCACCATGCCCGGCCATGTTTTATTTCTTTTTATT-TT

FAM87
Cen.ref GATTACAAGGTCAGGAGATCGAGACCATCCTGGCTAACACGGTGAAACCCTGTCTCTACTAAAGACACAAAAACAAAATTAGCCAGGTTTGGTGACGGGAGCCTG
BP      GATTACAAGGTCAGGAGATCGAGACCATCCTGGCTAACACGGTGAAACCCTGTCTCTACTAAAAATACAAA-----AATTAGGAGGGCATGGTGGCGCGTGCCTG
Tel.ref GATCACGAGGTCAAGAGTTCGAGACCAGCCTGGCCAACATAGTGAAACCCTGTCTCTACTAAAAATACAAA-----AATTAGGAGGGCATGGTGGCGCGTGCCTG

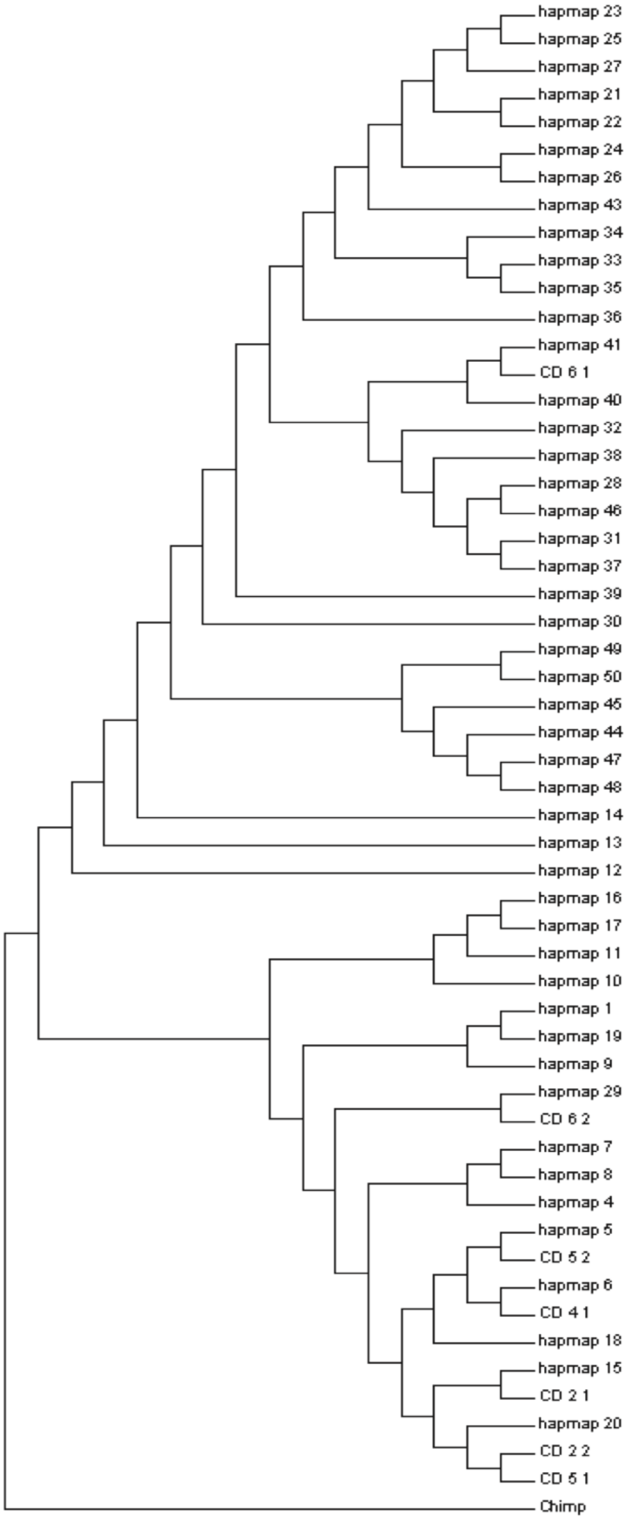
FAM23_d2
Cen.ref  AAAACAAAAAAAAACTGGAGGCCAGGCTCAGTGGGTCCCTCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGTGGATCACCTGAGGTCG
BP       AAAACAAAAAAAAACTGGAGGCCAGGCTCAGTGGGTCCCTCCTGTAATCCCAGCACTTTGGGAGGCCGAGGTGGGCAGATTACCTGAAGTCA
Tel.ref  -----AAAAAAAAAGCGTATG---------GTGGCGCGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGTGGGCAGATTACCTGAAGTCA


B.
FAM90, FAM91, FAM92, FAM93, FAM94 CD in FANCC
Cen.ref  AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-------------GTGAGCCGCGTTTTCCTAAGG-GCTGGCACACACAAAAAA-ACACTTTGTAGGCTGGG
BP FAM90 AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-AAAAAGTAAA-TTTAGTTAAAAGATTGCTTAATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT
BP FAM91 AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-AAAAAGTAAA-TTTAGTTAAAAGATTGCTTAATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT
BP FAM92 AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-AAAAAGTAAA-TTTAGTTAAAAGATTGCTTAATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT
BP FAM93 AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-AAAAAGTAAA-TTTAGTTAAAAGATTGCTTAATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT
BP FAM94 AGTATCTCTT--TTTCCCTGAC-----TGGAAGCTCCTAATGGGCA-GAGACA-AAAAAGTAAA-TTTAGTTAAAAGATTGCTTAATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT
Tel.ref  AGTTTCTCTTCATTTCTTAGAAAAAGTAAAATAATTTATTCCACATGTAACAATTTAAGCAATCTTTTAACTAAAGATTGCTTTATATGCTTGATCTCACTTCATTCACACGAAGAAACCATAT

FAM95
Cen.ref   GTTAGTATAAATACACAAATCAGTTAATAAATCATATTGATAAAGGAACAAAACCACAGTCATCTCAATAGATGCAGAAAAGCAAC-ATTTCTAACATTTGTTCATAAGAATT
BP        GTTAGTATAAATACACAAATCAGTTAATAAATCATATTGATAAAGGAACAC----ACTCCATTCCGACTAGGGGTGGCCA-GCAACTCAGCTGTAGCCAGTGAG-AGAGAAAG
Tel.ref   GTGCCCAATAAAAAGGACCACCTTCCCTACAAC-TAG------------------ACTCCATTCCGACTAGGGGTGGCCA-GCAACTCAGCTGTAGCCAGTGAG-AGAGAAAG
```

Supp. Figure S2. Alignment of sequences from breakpoint junctions to the reference genome. Alignments of the reference genomic sequence at the centromeric end of the breakpoint (Cen.ref) and the reference genomic sequence at the telomeric end of the breakpoint (Tel.ref) with the actual breakpoint junction sequence determined by sequencing (BP). Overlap sequence is underlined in red in the breakpoint sequence. Unaligned, inserted sequence is underlined in black in the breakpoint sequence. Yellow indicates sequence is identical in all, and blue indicates sequence is shared in two or more sequences. Alignments were done with AlignX, VNTI software (Life Technologies). **A.** *FANCA* breakpoint junctions. **B**. *FANCC* breakpoint junctions.

Supp. Figure S3A



Supp. Figure S3B

hapmap_1	AGTCGCGAGGGGGCTCGCTCCGCCCGGATTCCCCTATAAAAATCGG
hapmap_4	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGAATCGG
hapmap_5	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGAATTGG
hapmap_6	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGAATTGT
hapmap_7	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGAGCCAG
hapmap_8	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGGGCCAG
hapmap_9	AGTCGTGAGGGGGCTCGCTCCGCCCGGATTCCCCTGTAAAAATCGG
hapmap_10	AGTCGTGAGGGGGCTCGCTTCGCCTGGACTCCCTCGTAAAAATCGG
hapmap_11	AGTCGTGAGGGGGCTCGCTTCGTCTGGACTCCCTCGTAAAAATCGG
hapmap_12	AGTCGTGAGGGGGCTCGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_13	AGTCGTGAGGGGGCTCGTCCCGTCTAAGTCTTCTCATAGAAATCGG
hapmap_14	AGTCGTGAGGGGGCTCGTCCCGTCTAAGTCTTCTCGTAAAAATCGG
hapmap_15	AGTCGTGAGGGGGCTCGTTTCGTCTGGACTCCCCTGCGAGAATTGG
hapmap_16	AGTCGTGAGGGGGCTCGTTTCGTCTGGACTCCCTCGTAAAAATCGG
hapmap_17	AGTCGTGAGGGGGCTCGTTTCGTCTGGACTCCCTCGTAGAAATCGG
hapmap_18	AGTCGTGAGGGGGCTTGCTCCGCCCGGATTCCCCTGCGAGAATTGG
hapmap_19	AGTCGTGGGATGGCCCGCTCCGCCCGGATTCCCCTATAAAAATTGG
hapmap_20	AGTTGCGAGGGGGCTCGCTCCACCCGGATTCCCCTGCGAGAATTGG
hapmap_21	GTCCATAAAGGTATTTGTCCCGCCCAAGTCTCCTCATAAAAATTGG
hapmap_22	GTCCATAAAGGTATTTGTCCCGCCCAAGTTCCCTCATAAAAATTGG
hapmap_23	GTCCATAAAGGTATTTGTCCCGTCCAAGTCTCCTCATAAAAATTGG
hapmap_24	GTCCATAAAGGTATTTGTCCCGTCCAAGTCTCCTCATAAGAATTGT
hapmap_25	GTCCATAAAGGTATTTGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_26	GTCCATAAAGGTATTTGTCCCGTCCAGGTCCCCTCATAAGAATTGG
hapmap_27	GTCCATAAGGGTATTTGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_28	GTCCATGAAATGGCTCATTTCGTCTGGACTCCCTCGTAGAAATCGG
hapmap_29	GTCCATGAAGGTGCTCGCTCCACCCGGATTCCCCTGCGAAAATTGG
hapmap_30	GTCCATGAAGGTGCTCGTCCCGCCTAAGTCTTCTCATAAAAATCGG
hapmap_31	GTCCATGAAGGTGCTCGTTTCGCCTGGACCCCCTCGTAAGAGCCAG
hapmap_32	GTCCATGAAGGTGCTCGTTTCGTCTGGACCCCCTCGTAAAAATCGG
hapmap_33	GTCCATGAAGGTGTTTGTCCCGCCCAAGTCTTCTCATAAAAATTGG
hapmap_34	GTCCATGAAGGTGTTTGTCCCGTCCAAGTCTTCTCATAAAAATCGG
hapmap_35	GTCCATGAAGGTGTTTGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_36	GTCCATGAAGGTGTTTGTCCCGTCTAAGTCTTCTCGTAAGAATTGG
hapmap_37	GTCCATGAGGGTGCTCGTTTCGTCTGGACCCCCTCGTAAAAATCGG
hapmap_38	GTCCATGGAGGTGCTCGTTTCGTCTGGACCCCCTCGTAAAGATCGG
hapmap_39	GTCCATGGAGGTGTCCGTCCCGTCTAAGTCTTCTCATAGAAATCGG
hapmap_40	GTCCATGGAGGTGTTCGTCCTGTCCAAGTCTTCCTGCGAAGGTCAG
hapmap_41	GTCCATGGAGGTGTTTGTTTCGCCCGAGTTCTTCTGCGAGAGCCAG
hapmap_43	GTCCGCAAAGGTATTTGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_44	GTCCGCGGGATGGCCCGTCCCGCCCAAGTCTTCTCATAAAAATTGG
hapmap_45	GTCCGCGGGATGGCCCGTCCCGTCTAAGTCTTCTCATAGAAATCGG
hapmap_46	GTCCGCGGGATGGCCCGTTTCGTCTGGACTCCCTCATAAAAATTGG
hapmap_47	GTCCGCGGGATGGCTCGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_48	GTCCGCGGGATGGCTTGTCCCGTCCAAGTCTTCTCATAAAAATTGG
hapmap_49	GTCCGCGGGATGGTTTGTCCCGCCTAAGTCTTCTCATAGAAATCGG
hapmap_50	GTCCGCGGGATGGTTTGTCCCGTCTAAGTCTTCTCATAGAAATCGG
CD_2_1	AGTCGCGAGGGGGCTCGCTCCGTCCGGATTCCCCTGCGAGAATTGG
CD_2_2	AGTCGCGAGGGGGCTCGCTCCGCCCGGATTCCCCTGCGAGAATTGG
CD_4_1	AGTCGTGAGGGGGCCCGCTCCACCCGGATTCCCCTGCGAGAATTGT
CD_5_1	AGTTGCG?GGGGGCTCGC?CCGCCCGG?TT??C?TGCG?G??TTGG
CD_5_2	AGTCGTG?GGGGGCTCGC?CCGCCCGG?TT??C?TGCG?G??TTGG
CD_6_1	GTCCATGGAGGTGTTTGTTTCGCTTGAGTTCTTCTGCGAGAGCCAG
CD_6_2	AGTCGCGAGGGGGCTCGCTCCACCCGGATCCCCCTGCGAAAATCGG
Chimp	CCC?GTGGGGCGCGTG?ATGCGTCACAGTAGTACTCCGTTAGCCAG

Supp. Figure S3. Phylogenetic analysis of haplotypes from HAPMAP-CEU Phase 2 and FA patients with *FANCA* conserved deletions. **A**. A phylogenetic tree showing the predicted relationship of the haplotypes from *FANCA* common deletions with the unique haplotypes from the CEU population. **B**. The sequences used for the phylogenetics analysis. There are 55 total sequences: 47 are unique haplotypes from the CEU Hapmap population (Phase 2) (labeled "hapmap_*"), 7 are from the FA patients with *FANCA* common deletions (labeled "CD_*"), and 1 is from the Chimp genome which was used as an outgroup. Missing data was not evaluated (and marked with a "?").

Supp. Table S1. Array CGH - Targeted Genes and Their Chromosomal Region

Gene	Genomic Region (NCBI36/hg18)*
FANCA	chr16:88131460-88610566**
FANCB	chrX:14671450-14901105***
FANCC	chr9:96701501-97319867**
FANCD1 (BRCA2)	chr13:31777617-31881809
FANCD2	chr3:9993451-10166418****
FANCE	chr6:35518116-35552859
FANCF	chr11:22590655-22613963
FANCG	chr9:35013835-35120013****
FANCI	chr15:87578198-87671366
FANCJ (BRIP1)	chr17:57104767-57306537
FANCL	chr2:58229882-58333019
FANCM	chr14:44664886-44749843
FANCN (PALB2)	chr16:23511984-23570179
FANCO (RAD51C)	chr17:54114962-54176691
FANCP (SLX4) (BTBD12)	chr16:3561184-3611586
AP1TD1 (MHF1) (CENP-S)	chr1:10402746-10435459
BLM	chr15:89051583-89169690
BRCA1	chr17:38439840-38540994
CDKN2A	chr9:21947751-21994490
FAAP100 (C17orf70)	chr17:77107387-77139868
FAAP24 (C19orf40)	chr19:38144988-38169800
MRE11	chr11:93780115-93876688
NBN	chr8:91004740-91076075
RAD50	chr5:131910529-132017494
RAD51AP1	chr12:4508317-4549475
SMAD4	chr18:46800581-46875407
STRA13 (MHF2) (CENP-X)	chr17:77559868-77584062

* Target regions extend 10kb beyond either side of the gene, except

**Extends 200kb beyond either side of the gene

***Extends 100kb beyond either side of the gene

****Extends 50kb beyond either side of the gene

Supp. Table S2A. Additional Genes Affected in the Extended *FANCA* Deletions

Family ID	Δ Start	Δ End	Δ Size	Centromeric Genes Affected by Deletion
1	88,131,700	88,610,500	478,801	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A, DPEP1, CPNE7, RPL13, SPG7*
2	88,172,538	88,337,600	165,063	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A, DPEP1, CPNE7*
3	88,184,848	88,336,349	151,502	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A, DPEP1, CPNE7*
4	88,224,066	88,408,991	184,926	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A, DPEP1*
5	88,227,499	88,446,249	218,751	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A, DPEP1*
6	88,235,249	88,351,749	116,501	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A*
7	88,250,100	88,425,360	175,261	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55, CHMP1A*
8	88,257,445	88,409,236	151,792	*ZNF276, VSP9D1, SPATA2L, CDK10, C16orf55*
9	88,281,756	88,827,254	545,499	*ZNF276, VSP9D1, SPATA2L, CDK10*
10	88,285,490	88,362,122	76,633	*ZNF276, VSP9D1, SPATA2L, CDK10*
11	88,302,909	88,483,387	180,479	*ZNF276, VSP9D1*
12	88,311,999	88,394,999	83,001	*ZNF276, VSP9D1*
13	88,322,270	88,505,839	183,570	*ZNF276*
14	88,327,695	88,377,962	50,268	*ZNF276*
15	88,329,020	88,355,305	26,286	*ZNF276*
16	88,330,849	88,500,000	169,152	*ZNF276*
17	88,331,201	88,352,999	21,799	*ZNF276*
18	88,331,357	88,351,875	20,519	*ZNF276*
19	88,331,930	88,415,141	83,212	*ZNF276*
20	88,334,499	88,393,249	58,751	*ZNF276*

Family ID	Δ Start	Δ End	Δ Size	Telomeric Genes Affected by Deletion
88	88,409,549	88,411,749	2,201	*none; FANCA-SPIRE2 intergenic*
65	88,387,000	88,412,000	25,001	*none; FANCA-SPIRE2 intergenic*
72	88,399,249	88,412,249	13,001	*none; FANCA-SPIRE2 intergenic*
21	88,336,484	88,414,112	77,629	*none; FANCA-SPIRE2 intergenic*
23_d2	88,406,326	88,414,712	8,387	*none; FANCA-SPIRE2 intergenic*
19	88,331,930	88,415,141	83,212	*none; FANCA-SPIRE2 intergenic*
83	88,403,104	88,418,467	15,364	*none; FANCA-SPIRE2 intergenic*
82	88,403,094	88,418,774	15,681	*none; FANCA-SPIRE2 intergenic*
81	88,403,094	88,418,774	15,681	*none; FANCA-SPIRE2 intergenic*
74	88,400,101	88,419,559	19,459	*none; FANCA-SPIRE2 intergenic*
69	88,398,749	88,420,249	21,501	*none; FANCA-SPIRE2 intergenic*
7	88,250,100	88,425,360	175,261	*SPIRE2*
70	88,398,749	88,425,749	27,001	*SPIRE2*
66	88,387,749	88,432,749	45,001	*SPIRE2*
75	88,401,663	88,433,153	31,491	*SPIRE2*
76	88,401,663	88,433,153	31,491	*SPIRE2*

77	88,401,663	88,433,153	31,491	*SPIRE2*
78	88,401,663	88,433,153	31,491	*SPIRE2*
79	88,401,663	88,433,153	31,491	*SPIRE2*
29	88,347,917	88,433,278	85,362	*SPIRE2*
80	88,402,939	88,435,730	32,792	*SPIRE2*
85	88,403,999	88,437,249	33,251	*SPIRE2*
62	88,383,499	88,437,499	54,001	*SPIRE2*
5	88,227,499	88,446,249	218,751	*SPIRE2*
28_d1	88,345,766	88,460,603	114,838	*SPIRE2*
22	88,338,249	88,470,249	132,001	*SPIRE2, TCF25*
61	88,383,499	88,474,299	90,801	*SPIRE2, TCF25*
11	88,302,909	88,483,387	180,479	*SPIRE2, TCF25*
16	88,330,849	88,500,000	169,152	*SPIRE2, TCF25*
13	88,322,270	88,505,839	183,570	*SPIRE2, TCF25*
1	88,131,700	88,610,500	478,801	*SPIRE2, TCF25, MC1R, TUBB3, DEF8, CENPBD1, AFG3L1P, DBNDD1*
9	88,281,756	88,827,254	545,499	*SPIRE2, TCF25, MC1R, TUBB3, DEF8, CENPBD1, AFG3L1P, DBNDD1, GAS81,C16ORF3, LOC100130015, PRMD7*

Supp. Table S2B. Description of the Neighboring Genes in the Extended FANCA Deletions

Location Relative to FANCA	Gene	RefSeq_ID	Location	Gene Product
Centromeric	*SPG7*	NM_003119.2	chr16:88102306-88151675	paraplegin isoform 1 precursor
	RPL13	NM_001243131.1	chr16:88154566-88160738	60S ribosomal protein L13 isoform 3
	CPNE7	NM_153636.2	chr16:88169677-88191155	copine-7 isoform a
	DPEP1	NM_001128141.2	chr16:88207217-88232340	dipeptidase 1 precursor
	CHMP1A	NM_002768.3	chr16:88238340-88251630	charged multivesicular body protein 1a isoform 2
	C16orf55	NM_153025.1	chr16:88251711-88265176	uncharacterized protein C16orf55
	CDK10	NM_001160367.1	chr16:88280577-88290273	cyclin-dependent kinase 10 isoform d
	SPATA2L	NM_152339.3	chr16:88290266-88295622	spermatogenesis-associated protein 2-like protein
	VSP9D1	NM_004913.2	chr16:88301042-88314895	Unknown protein
	ZNF276	NM_001113525.1	chr16:88315453-88334833	zinc finger protein 276 isoform a
Telomeric	*SPIRE2*	NM_032451.1	chr16:88422408-88465228	protein spire homolog 2
	TCF25	NM_014972.2	chr16:88467495-88505293	transcription factor 25
	MC1R	NM_002386.3	chr16:88511788-88514886	melanocyte-stimulating hormone receptor
	TUBB3	NM_006086	chr16:88517188-88530006	PubMed on Product: tubulin beta-3 chain isoform 1
	DEF8	NM_001242819.1	chr16:88543329-88561969	differentially expressed in FDCP 8 homolog isoform 6
	CENPBD1	NM_145039.3	chr16:88563684-88566741	CENPB DNA-binding domains containing 1
	AFG3L1P	NR_003226.1	chr16:88566489-88590529	pseudogene AFG3 ATPase family gene 3-like 1 (AFG3L1P)
	DBNDD1	NM_001042610.1	chr16:88598780-88613438	dysbindin domain containing 1 (DBNDD1)
	GAS8	NM_001481.2	chr16:88616509-88638880	Homo sapiens growth arrest-specific 8 (GAS8),
	C16orf3	NM_001214.3	chr16:88622817-88623810	Homo sapiens chromosome 16 open reading frame 3
	LOC100130015	NR_027335.2	chr16:88633670-88641534	Homo sapiens 5-hydroxyisourate hydrolase pseudogene
	PRDM7	NM_001098173.1	chr16:88650475-88669839	Homo sapiens PR domain containing 7 (PRDM7)

The genomic coordinates are according to the NCBI36/hg18 build.

Supp. Table S3. Primer Sequences Used for Cloning and Sequencing Deletion Breakpoints

FAM ID	Sense Primer	Antisense Primer	Size (bp)	FA Gene
2	CATATCAGCACCTAAAAGTCTCCTGGT	CATCCTGAGTTCCCAGAACCGTC	2894	*FANCA*
4	GTAGCTGGAGTGTGGGTCCCTGTGA	GAGATCACGCCCCTTGCAATC	735	*FANCA*
7	GCGGGAGGTAGATGTTATCTTTCATC	CCTTCACATGGGCCGTAACCA	881	*FANCA*
8	TCTCGGGTCTCAACTCACTGTAGCC	AAGGCCATGTCCGACTCGTG	2344	*FANCA*
10	TCGCCCCAATTCTCCCTGAGGT	GGGCCATCCTCTCTTAGAAACACAG	889	*FANCA*
11	CGCACAGACACAGATGATCCGC	TCCCGAAGAAACCCATCAACAC	1193	*FANCA*
13	CTTCAAAAACCATCAGTGACCTTTCTG	TGGGGCTTCGCCTTCAATTCTGG	1188	*FANCA*
14	CAAGTGTCCTTACCAGGGCTGCACG	GTGAGTGCGTGTGTGCCAGTCAAGG	532	*FANCA*
15	GACGCACACTGGAAGCTGCTGCAT	CACGCGGTGGCAATTTCTGTTT	2375	*FANCA*
17	TAGAGAACAGCCATAGCTGTGACAATCAGT	ATATGGAGTATCCTGGTGGGTATATGGG	1573	*FANCA*
18	TAGAGAACAGCCATAGCTGTGACAATCAGT	CCATCACACACCGATGAGGCCTT	505	*FANCA*
19	TGGGGCTGGTCTTGAACTGACTTC	TTGTAAGATGAGAGAACTAGGCTGGACGT	1902	*FANCA*
21	TGTAAGTTGTGTGCTGACATGGCA	TGCCAGATCAAAGGACAGATGAACT	2021	*FANCA*
23_d1	GCTGTGGATTCAGTTTTGAGAAAA	GGTTCGTCATGTTTTTCTCTGAATCAC	1375	*FANCA*
24	CTGCGGAAAATCTCAAAGAGGAAGTGC	AACTCCCCGGGCAGTGTTGTT	779	*FANCA*
25	CCTTGAGCCCAGGAGTTACAGTACTAC	ACTTAGCTGGTCATGGTGAT	1803	*FANCA*
26	TTATCAATTAAGAAAAAACAGCTGGGCG	CACGTTTAGCCTTGGGTATATTAGTATGG	732	*FANCA*
27	CCTTGAGCCCAGGAGTTACAGTACTAC	GCGCCTGGCCTAATATTTTGG	2616	*FANCA*
28_d1	TCGAGCTCAGCTGATTCTCCTAAGTC	TGATGGGTCACATACTTTTGGAAAGTT	1366	*FANCA*
29	TCAACACTGCACTGGGGACAGACA	GGCTGCCCTGCCTTTGACTCTCTTA	942	*FANCA*
30	ACTCAACACTGCACTGGGGACAGA	GTATAGAGGGGAATAAACAAGCCTGCC	1710	*FANCA*
31	GCAGTGAGCAGCTCTGTGCAAACC	GCTGCCTTTCCTTGTGTTCTCTGAG	2006	*FANCA*
32	TCAAGGCTGGCATCACTCCTCA	AGCCTTCCGGTGATTCAATGATG	1601	*FANCA*
33	CTCCAGGTGTAACCAGTCTTGGTAAGT	TTCTAGATCCCCTTCCACCTGCA	1190	*FANCA*
34	GAAAGTTCCCTCTTACTAATTTGCTAAGGA	CTGAGAAAGAGAGCAAGCAATTCAATAC	1310	*FANCA*
35	CTGAGAAAGAGAGCAAGCAATTCAATAC	TCCCCTGTCGTGCTGGTATTTTG	1784	*FANCA*
36	CCAAAACAACCAGGCAAACATCCTC	GGTTCGTCATGTTTTTCTCTGAATCAC	624	*FANCA*
37	CCAAAACAACCAGGCAAACATCCTC	GGTTCGTCATGTTTTTCTCTGAATCAC	624	*FANCA*
38	CCAAAACAACCAGGCAAACATCCTC	GGTTCGTCATGTTTTTCTCTGAATCAC	624	*FANCA*
39	TAACCAGGCACACATGGGTGAG	AGCCTTCCGGTGATTCAATGATG	1241	*FANCA*
40	TAACCAGGCACACATGGGTGAG	TCCTGTGGGGTTGAATGGAGAGG	747	*FANCA*
44	TTGCCTCCATGCGGAGCTTG	ATTCTCACCAGCATTTGTTA	838	*FANCA*
45	GCGCACTTGGTCTGAGTGAAGAC	GAGTTCTGTTTGCATTCTCACCAGC	923	*FANCA*
46_d1	TCTCCTCTTAGTTTAGTGGAAGCCCC	CACCTGGCACAAGCCTATAGTTTTG	1046	*FANCA*
47_d1	GGAGGACACCTCTCAAGAGAAGCA	TATAAGAGTTCTGTTTGCATTCTCACCAGC	1893	*FANCA*
48	ATCTGACTGCTAATGGAGTTTCATGCCA	GGGAGCATTTTACCTGTTAATTATGGTT	907	*FANCA*
49	GGAGGACACCTCTCAAGAGAAGCA	ATGGCCGGCCTAATGAAAACA	442	*FANCA*
50	AGAGGTCATGCTCTCAGGGTGGGTC	TCCCCTGTCGTGCTGGTATTTTG	941	*FANCA*
51	AGAGGTCATGCTCTCAGGGTGGGTC	TCCCCTGTCGTGCTGGTATTTTG	941	*FANCA*
52	TGAAGAATGATGCAGCCGGGTGC	GTTCTACTCTCTACCTCCACAAGAGCCACT	777	*FANCA*
53	TCATCATGCTACCCTACTGCAGCC	GGGAAGCAGCCTTCTATTGTGTTTT	1142	*FANCA*
54	TCATCATGCTACCCTACTGCAGCC	CCAGAGTGTCGAAGTGTTGGGATT	1214	*FANCA*

55	ACCATCTGACATTTGGCTGCTATAAA	GATTGCACATCCCTGGTTCCCC	451	*FANCA*
56	ACCATCTGACATTTGGCTGCTATAAA	GGGAAGCAGCCTTCTATTGTGTTTT	369	*FANCA*
57	ACCATCTGACATTTGGCTGCTATAAA	GCACCTTAACCCCCGAAGTACTGG	567	*FANCA*
46_d2	GGAGGACACCTCTCAAGAGAAGCA	CACGTTTAGCCTTGGGTATATTAGTATGG	528	*FANCA*
63_d2	TCTCCTCTTAGTTTAGTGGAAGCCCC	CACCTGGCACAAGCCTATAGTTTTG	1046	*FANCA*
58	TCTCCTCTTAGTTTAGTGGAAGCCCC	CACCTGGCACAAGCCTATAGTTTTG	1046	*FANCA*
59	TCTCCTCTTAGTTTAGTGGAAGCCCC	CACCTGGCACAAGCCTATAGTTTTG	1046	*FANCA*
60	AGAGGTCATGCTCTCAGGGTGGGTC	AGAGGCCAGAGTGTCCACTGCTGCT	1653	*FANCA*
47_d2	TTTCAGGACTTACGTGAAAATTAGAGACG	CCTTTCCTTTGGCTGTAAGCCCAG	1568	*FANCA*
28_d2	TCTCGTGAGGCTTACAACCTGCA	GAGCCAGTACATTACAATAACAGGTTCG	735	*FANCA*
67	AAGAAACGGCACTTCAGGAGGC	CGGGTTTGATTTCTGGCTTTGAC	646	*FANCA*
68	AACATACACACAGACCAACGGAACAGG	CGTTTAGCCTTGGGTATATTAGTATGGAG	467	*FANCA*
71	TCCTCAGGAAGGCCAGGCATAGT	GAGCCAGTACATTACAATAACAGGTTCG	857	*FANCA*
73	TTCCCAATTCCCTTCTGTTGTTTATTC	ATGCCCTTTTGCTTGAGGTAAGAGGA	967	*FANCA*
74	CCCAATTCCCTTCTGTTGTTTATTCT	CGAATTGTGCCTAAAGTTTGTTTGTTTG	1507	*FANCA*
75	CCAAGGCAGAAGAATTTCTCTTAGTACAG	TGTGAGTGAAGCCATCTCTGATCTCT	1468	*FANCA*
76	CAGAACAAAATGAAAAGTCTCCCGTG	TTGACTCTCTTAGCCTTGGGGTCAG	1627	*FANCA*
77	AAATTCTGCTCTAGGCCGAGGC	GGCTGCCCTGCCTTTGACTCTCTTA	1151	*FANCA*
78	CCAAGGCAGAAGAATTTCTCTTAGTACAG	TGTGAGTGAAGCCATCTCTGATCTCT	1469	*FANCA*
79	AAATTCTGCTCTAGGCCGAGGC	GGCTGCCCTGCCTTTGACTCTCTTA	1151	*FANCA*
80	CACTGTGAACACTGTGCCTGTCA	GCCCAGGATGTTATAAAACTCAGATGT	460	*FANCA*
81	GCACTGTAGCCTACTTCTAACCCTTTCT	TCTTCCTCAAAGGGCCTCACGT	843	*FANCA*
82	GCACTGTAGCCTACTTCTAACCCTTTCT	TCTTCCTCAAAGGGCCTCACGT	843	*FANCA*
83	GCACTGTAGCCTACTTCTAACCCTTTCT	TCTTCCTCAAAGGGCCTCACGT	1159	*FANCA*
87	CCCAGAGTTATACATTGGTATTTGTTACGT	ATGCCCTTTTGCTTGAGGTAAGAGGA	2002	*FANCA*
23_d2	CCCAGAGTTATACATTGGTATTTGTTACGT	TGCCAGATCAAAGGACAGATGAACTT	1413	*FANCA*
90	ATCTGACTGCTAATGGAGTTTCATGCCA	GGGAGCATTTTACCTGTTAATTATGGTT	907	*FANCC*
91	ATCTGACTGCTAATGGAGTTTCATGCCA	GGGAGCATTTTACCTGTTAATTATGGTT	907	*FANCC*
92	ATCTGACTGCTAATGGAGTTTCATGCCA	GGGAGCATTTTACCTGTTAATTATGGTT	907	*FANCC*
93	ATCTGACTGCTAATGGAGTTTCATGCCA	GGGAGCATTTTACCTGTTAATTATGGTT	907	*FANCC*
94	CTGTGCAGCCTCAGCTCCTCAGAA	ACTGAACCGCCAGGACCCTTGATT	515	*FANCC*
95	GTATGTGGAAGCTGGCGATGGA	ACCCAGGGTCCTCCTTCTTCTTT	680	*FANCC*

Supp. Table S4. Overlap or Insertion Sequences at Deletion Breakpoints in FA Families

FAM ID	Overlap or Insertion Sequences*
2	GGCATGGTGGC
4	CAG
7	AGTCTCGCTCTGTCGCCCAGGCTGGAGTG
8	GCTAGGATTACAGGCGTGAGCCACCA
10	AG
11	CCAG
13	GCCTCAGCCTCCCGAGTAGCTGGG
14	GC
15	TCCAGCCTGGG
17	GAGG; a SNP near overlap
18	GG
19	AG
21	GTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGC
23_d1	ACTTTGGGAGGCCAAGGCGGG
24	CCAGCACTTTGGGAGGCCGAGGCAGGCAGAT
25	GAGACCA
26	TGCAGTGAGCCAAGATC
27	CCTGTAATCC
28_d1	CCAGGCTGGAGTGCAGTGGCACAATCTCGGCTCACTGCAAGCTCC
29	AAAAAAAAAAA
30	*ins*. ttccagcttccccagcctttatgggtaggtttgcact
31	*ins*. tggagcctgaCCCAGCCTgacccagcctg
32	AC
33	GCTAACACGGTGAAACCCTGT
34	GAGGTCAGGAGATTGAGACCATCCTGGCTAACACAGTGAAACCC
35	*ins*.cacacaaaattagcctgacgtggtggcgggtgcctgtagtcccagctactcgggaggctgaggcaggagaatggcatgaacccggaaggcggagcttgcagtgagctgagactgcgccactgcactccagcctgggcaatagaacgagactccattccaaaaaaaaaaaaaaaaacaag
36	AAAA
37	AAAA
38	AAAA
39	CGCCTGTAGTCCCAGCTACT
40	GAGGCCAAGGCG
44	GCTTGAACCC
45	GCTTGAACCC
46_d1	CTGGCTAACACGGTGAAACCCCGTCTCT
47_d1	GTCTCTACTAAAAA
48	CGCCTGTA
49	GGGAGGCGGAGCTT
50	AC

51	AC
52	none
53	CCTGTAATCCCAG
54	GAGGCTGAGGCAGGAGAATGGT
55	TAATCCCAGCACTTTGGGAGGC
56	GTCTCTACTAAAAA
57	ATTAGCCAGG
46_d2	*ins*. tt
63_d2	*ins*. tt
58	*ins*. tt
59	*ins*. tt
60	*ins*. tt
47_d2	CTGAGGCAGGAGAATAGCTTGAA
28_d2	TGGGAGGC
67	GTGGCGGACGCC
68	TAGTCCCAGCTACTCGGGAG
71	A
73	CTCAC
74	GCCTCCCAAAGTGCTGGGATTACAGG
75	GGCGTGGTGGCGG
76	GGCGTGGTGGCGG
77	GGCGTGGTGGCGG
78	GGCGTGGTGGCGG
79	GGCGTGGTGGCGG
80	TGCCTCAGCCTCC
81	GGCCTCCCAAAGTGCTGGGATTACAGG
82	GGCCTCCCAAAGTGCTGGGATTACAGG
83	CCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCAC
87	GTGAAACCCTGTCTCTACTAAA
23_d2	CCTGTAATCCCAGCACTTTGGGAGGCCGAGG
90	*ins*. aaaaagtaaatttagtta
91	*ins*. aaaaagtaaatttagtta
92	*ins*. aaaaagtaaatttagtta
93	*ins*. aaaaagtaaatttagtta
94	*ins*. aaaaagtaaatttagtta
95	*ins*. c

Details of the characteristics of the breakpoints are provided in Table 3.

*Overlap sequence is capitalized and insertion sequence is in lower case and begins prefix "*ins.*"

Identical deletions are shaded in gray.

Supp. Table S5. Families With Conserved Deletions in FANCA and FANCC and Their Ancestry

Gene	Common Deletions (CD)	Exons	Family ID	Ancestry
FANCA	(CD1) chr16:88357692-88376135	exons 18-28	36	European/Caucasian
			37	European/Caucasian
			38	European/Caucasian
FANCA	(CD2) chr16:88366653-88394866	exons 9-22	44	European/Caucasian
			45	European/Caucasian
FANCA	(CD3) chr16:88368840-88373628	exons 18-21	50	European/Caucasian
			51	French, Hispanic/Caucasian
FANCA	(CD4) chr:88383475-88391064	exons 11-14	46_d2	European-American Indian/ Caucasian
			58	European/Caucasian
			59	European/Caucasian
			60	European/Caucasian
			63_d2	African-American
FANCA	(CD5) chr16:88401663-88433153	exons 1-6	75	European/Caucasian
			76	European/Caucasian
			77	European/Caucasian
			78	European/Caucasian
			79	European/Caucasian
FANCA	(CD6) chr16:88403094-88418774	exons 1-5	81	European/Caucasian
			82	Hispanic
			83*	European/Caucasian
FANCC	chr9:97046452-97050240	exon 3	90	European/Caucasian
			91	European/Caucasian
			92	European/Caucasian
			93	European/Caucasian
			94	European/Caucasian

*deletion chr16:88403104-88418467

The genomic coordinates are according to the NCBI36/hg18 build.

Supp. Table S6. *Alu* Elements in *FANCA* with Multiple Breakpoints (BP)

SINE Element	Start	End	Intron	Unique BP	CDBP
*Alu*Sx	88,347,682	88,347,981	30	2	-
*Alu*Y	88,355,047	88,355,357	29	2	-
*Alu*Y	88,356,875	88,357,188	28	3	-
*Alu*Sg	88,357,566	88,357,859	28	2	CD1_CBP
*Alu*Y	88,368,358	88,368,663	21	5	-
*Alu*Y	88,375,302	88,375,614	17	4	-
*Alu*Y	88,376,003	88,376,138	17	2	CD1_TBP
*Alu*Y	88,376,139	88,376,434	17	2	-
*Alu*Y	88,377,178	88,377,485	15	3	-
*Alu*Y	88,393,654	88,393,920	8	2	-
*Alu*Sx	88,394,658	88,394,971	8	3	CD2_TBP
*Alu*Sq	88,399,826	88,400,121	6	2	-
*Alu*Y	88,401,513	88,401,817	6	2	CD5_CBP
*Alu*Y	88,402,820	88,403,114	5	4	CD6_CBP
*Alu*Y	88,403,951	88,404,249	5	2	-
*Alu*Y	88,405,963	88,406,268	3	2	-
*Alu*Sp	88,406,274	88,406,579	3	2	-
*Alu*Sx	88,408,947	88,409,245	2	3	-
*Alu*Sg	88,418,482	88,418,795	downstream	2	CD6_TBP
*Alu*Y	88,432,969	88,433,298	downstream	2	CD5_TBP

CDBP = Conserved Deletion Breakpoint
CBP = Centromeric Breakpoint
TBP = Telomeric Breakpoint
The chr 16 genomic coordinates are according to the NCBI36/hg18 build.