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Genome-Wide Structural Variation Detection by Genome Mapping on Nanochannel Arrays

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ABSTRACT Comprehensive whole-genome structural variation detection is challenging with current approaches. With diploid cells as DNA source and the presence of numerous repetitive elements, short-read DNA sequencing cannot be used to detect structural variation efficiently. In this report, we show that genome mapping with long, fluorescently labeled DNA molecules imaged on nanochannel arrays can be used for whole-genome structural variation detection without sequencing. While whole-genome haplotyping is not achieved, local phasing (across >150-kb regions) is routine, as molecules from the parental chromosomes are examined separately. In one experiment, we generated genome maps from a trio from the 1000 Genomes Project, compared the maps against that derived from the reference human genome, and identified structural variations that are >5 kb in size. We find that these individuals have many more structural variants than those published, including some with the potential of disrupting gene function or regulation.

KEYWORDS biotechnology; genome mapping; structural variation detection

WHOLE-GENOME short-read sequencing is now routine and affordable. However, three challenges remain in genome analysis: genome sequence assembly, structural variation detection, and separation of the two parental genomes. In addition to the fact that humans are diploid, with cells harboring two genomes from the parents, the presence of numerous repetitive elements that are longer than the usual sequencing library insert size makes it close to impossible to assemble genome sequences with short-read sequencing alone (El-Metwally *et al.* 2013). Consequently, almost all whole-genome sequencing projects map the sequencing reads onto the human reference genome sequence without performing whole-genome assemblies (Ley *et al.* 2008). When whole-genome assembly is attempted, it is done by the laborious and expensive approach of generating paired-end sequencing of cloned genomic DNA fragments to provide scaffolds for sequence assembly (Siegel *et al.* 2000). Alignment of short sequencing reads to the human reference genome sequence reveals single-nucleotide variation and small indels in the individuals sequenced, but larger structural variants and repetitive regions in the genome are more difficult to detect. As structural variation can disrupt genes or regulatory elements, whole-genome sequencing without assembly and detection of structural variation produces an incomplete picture of the genome. Recently, clone-free approaches (*e.g.*, Hi-C scaffolding) have been used to generate sequence motif maps or long sequences to serve as scaffolds for the assembly of highly accurate short-read sequences (Burton *et al.* 2013; Kaplan and Dekker 2013), including the

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de novo assembly of a diploid human genome (Pendleton *et al.* 2015). These "hybrid assembly" approaches rely on three sets of data—short read sequences, long read sequences (5- to 20-kb reads), and genome maps (150–500 kb)—to overcome repetitive elements and duplicated regions larger than the typical contigs assembled from short-read sequences.

A fully assembled and phased diploid genome makes it possible to identify all structural variants present with direct access to the breakpoints involved. However, high-quality human genome sequence assembly with base-pair resolution, while feasible, is still a costly and laborious endeavor. In this report, we demonstrate the utility of genome mapping, an approach based on massively parallel analysis of extremely long single DNA molecules fluorescently labeled at specific sequence motifs in nanochannel arrays in genome-wide identification of structural variation at 5-kb resolution without sequencing. In contrast to the short reads (hundreds of bases) used in next-generation sequencing (NGS) approaches, genome mapping analyzes individual DNA molecules of hundreds of thousands of base pairs, thus preserving the long-range genome architecture and enabling direct interrogation of structural variants. Genome mapping has been used in several previous studies to provide scaffolds for genome sequence assembly (Hastie *et al.* 2013; Cao *et al.* 2014; English *et al.* 2015; Pendleton *et al.* 2015; Usher *et al.* 2015; Xiao *et al.* 2015). The DNA sample is prepared with a protocol that preserves the integrity of the DNA. Because native DNA is used, no amplification bias is present. Currently, analyzing a genome by genome mapping (at $>60\times$ coverage) takes <2 days and costs $<\$1000$. While whole-genome haplotyping is not achieved with genome mapping alone, local phasing across regions of at least 150 kb is routine with our single-molecule analysis approach, as molecules derived from the parental chromosomes are examined separately.

We generated genome maps from a trio from the 1000 Genomes Project where the individuals had been sequenced to high depths and with their structural variations published previously. We compared the genome maps obtained from the trio against those derived from the human reference genome and identified all the structural variations that are >5 kb. Comparing the genome maps of the parents and the child allows us to check for consistency in Mendelian inheritance and separate the haplotypes. Our study shows that these individuals have many more structural variants than those published and that some of these variants have the potential to disrupt gene function or regulation. Using nicking endonuclease Nt.BspQI (GCTCTTCN^), one label is observed about every 8 kb in the human genome. Without sequencing or cloning, we are able to map breakpoints of the structural variation within 8 kb, making this a novel and efficient approach to whole-genome structural variation analysis. Furthermore, our maps pinpoint the Epstein–Barr Virus (EBV) integration sites in the lymphoblastoid cell lines used and provide size estimates of two-thirds of the large "N-base gaps" in the hg38 human reference genome sequence.

Materials and Methods

High-molecular-weight DNA extraction

Cells from the trio cell line were washed with PBS, resuspended in cell suspension buffer, and embedded in thin low-melting-point agarose layers (CHEF Genomic DNA Plug Kit, Bio-Rad). The thin agarose layers were incubated with lysis buffer and proteinase K for 4 hr at 50°. The plugs were washed and then solubilized with GELase (Epicentre). The purified DNA was subjected to 4 hr of drop-dialysis. It was quantified using Nanodrop 1000 (Thermal Fisher Scientific) and/or a Quant-iTdsDNA Assay Kit (Invitrogen/Molecular Probes), and the quality was assessed using pulsed-field gel electrophoresis.

DNA labeling

The DNA was labeled according to commercial protocols using the IrysPrep Reagent Kit (BioNano Genomics). Specifically, 300 ng of purified genomic DNA was nicked with 7 U of nicking endonuclease Nt.BspQI [New England BioLabs (NEB)] at 37° for 2 hr in NEB Buffer 3. The nicked DNA was labeled with a fluorescent-dUTP nucleotide analog using Taq polymerare (NEB) for 1 hr at 72°. After labeling, the nicks were ligated with Taq ligase (NEB) in the presence of dNTPs. The backbone of fluorescently labeled DNA was counterstained with YOYO-1 (Invitrogen).

Data collection

The DNA was loaded onto the nanochannel array of BioNano Genomics IrysChip by electrophoresis of DNA, automated by the Irys system. Linearized DNA molecules were imaged using the BioNano Genomics Irys system. The DNA backbone (outlined by YOYO-1 staining) and locations of fluorescent labels along each molecule were detected using the image detection software of the Irys system, IrysView, which is available upon request (http://www.bionanogenomics.com/products/irysview/). The set of label locations of each DNA molecule defines an individual single-molecule map.

De novo genome map assembly

Single-molecule maps were assembled *de novo* into genome maps using IrysSolve software tools developed at BioNano Genomics (available upon request) (Cao *et al.* 2014). Briefly, the assembler is a custom implementation of the overlap-layout-consensus paradigm with a maximum-likelihood model. An overlap graph was generated based on pairwise comparison of all molecules as input. Redundant and spurious edges were removed. The assembler output the longest path in the graph, and consensus maps were derived. Consensus maps were further refined by mapping single-molecule maps to consensus maps, and label positions were recalculated. Refined consensus maps were extended by mapping single molecules to the ends of the consensus and calculating label positions beyond the initial maps. After merging of overlapping maps, a final set of consensus maps (genome maps) was output and used for subsequent analysis.

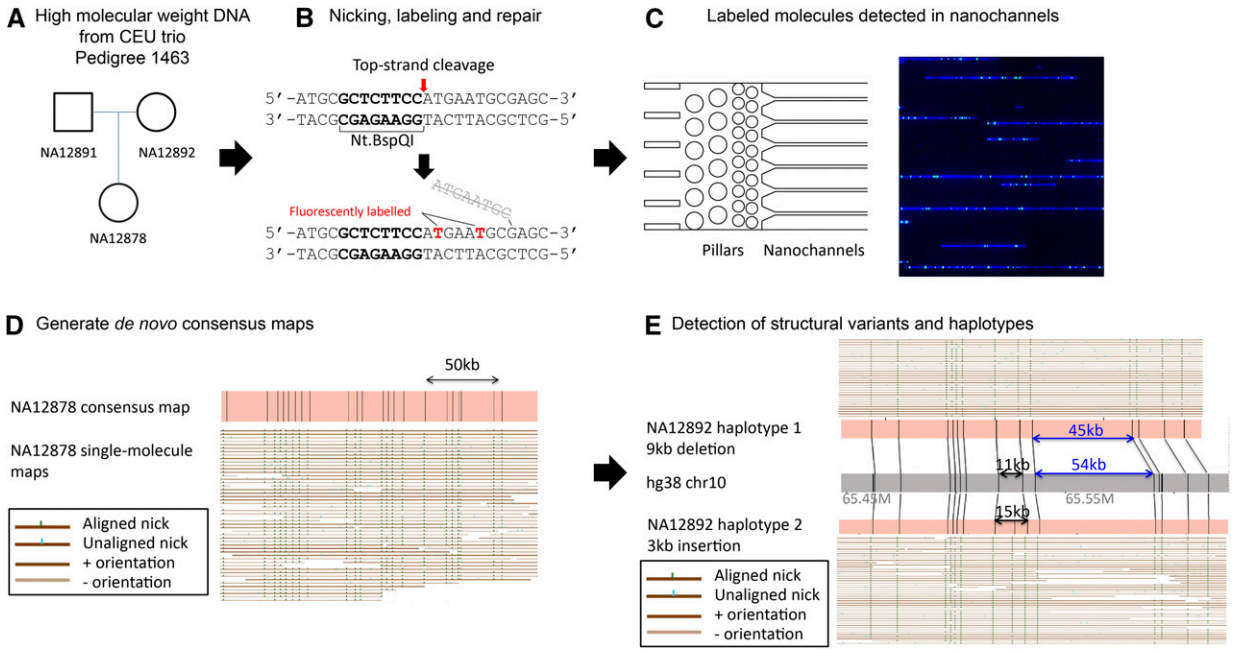

Figure 1 Overview of genome mapping strategy. (A) High-molecular-weight DNA was extracted from cell culture of the trio cell lines. (B) Nt.BspQI nicking endonuclease was used to nick the top strand of the DNA. The top strand was then displaced with fluorescently labeled thymine using *Taq* DNA polymerase. The displaced strand was simultaneously removed by the 5′ flap endonuclease activity of *Taq* DNA polymerase. The nicked DNA was then repaired by *Taq* ligase. The DNA backbone was fluorescently stained by YOYO-1. (C) Labeled DNA molecules were loaded onto flowcells where they uncoiled in the gradient pillar region before they entered the nanochannels where they were imaged. Molecule size and BspQI label locations were determined to generate single-molecule maps. (D) Single-molecule maps were assembled *de novo* into genome maps. (E) Genome maps were compared with hg38 *in silico* maps to detect structural variants and identify heterozygous regions.

Generation of in silico BspQI maps

EMBOSS restrict (Rice *et al.* 2000) was used to detect *in silico* BspQI sites and generate maps for the hg38 reference genome and the accompanied EBV genome. To match the resolution of the *in silico* map, which was originally at a 1-bp resolution, to the experimental data, which was at an ~500-bp resolution, the *in silico* maps were condensed to 700 bp such that a midpoint position would be taken if two or more BspQI sites were within 700 bp of each other.

Identification EBV integration sites

Two tandem copies of the EBV *in silico* BspQI maps were used as the EBV *in silico* reference map to account for the circular nature of the EBV genome and prevent mapping artifacts due to a linearized map. Single-molecule maps were aligned to the EBV *in silico* reference map using software tools developed at BioNano Genomics (Shelton *et al.* 2015). For single-molecule maps that were partially aligned to the EBV reference maps, unmapped portions that were >100 kb and had a minimum of 10 nicks were mapped to hg38 to determine potential EBV integration sites. High-confidence EBV integration sites were supported by at least 20 single-molecule maps.

Sizing N-base gaps

N-base gaps in hg38 that were >5 kb were identified and those that corresponded to centromeres and telomeres were removed. Centromere and telomere annotations were based on hg38 centromeres and gap annotations on the University of California at Santa Cruz (UCSC) Table Browser. Using alignment of genome maps to the hg38 *in silico* BspQI map (described below), genome maps that span across the entire N-base gaps were used to estimate the size of the N-base gaps.

Structural variation detection

Structural variants (SVs) were found by identifying outlier alignments between single-molecule maps or genome maps from a sample and the hg38 reference map. A structural variation detection pipeline that takes advantage of three detection strategies was used to detect SVs (see below). SVs were verified manually to confirm supporting evidence from single-molecule maps. Briefly, each of the >1000 SVs identified by the software tools was checked by two expert analysts to confirm that (1) at least 50 high-quality single-molecule maps covered the SV region and (2) there was clear evidence that a deletion or an insertion event was found in at least 20% of the high-quality single-molecule maps (for a heterozygous SV call) or in >90% of the high-quality single-molecule maps (for a homozygous SV call).

Structural variation detection pipeline

The first SV list was generated using BioNano software tools. Structural variants were found by identifying outliers between genome maps and the hg38 reference *in silico* map. An outlier is defined as a discrepancy between the two maps at the

0.01% level or worse. Outliers that are found between two high-scoring regions of at least 50 kb are considered SVs.

Two other software tools are also used to identify SVs based on alignment of single-molecule maps directly to the hg38 reference map. A pipeline was set up to apply these two methods for SV detection. Each single-molecule map was aligned to the reference maps by two different alignment algorithms. The alignment results were integrated and supplied as inputs to two SV calling methods that used different approaches to identify SVs. Detailed descriptions of the pipeline are in unpublished results (K. Y. Yip *et al.* and T. F. Chan *et al.*). Below are the key steps of this pipeline.

Alignment of single-molecule maps to reference maps: To align single-molecule maps to the hg38 reference maps, we used RefAligner (BioNano Genomics) (Cao *et al.* 2014) and OMBlast (Leung *et al.*, unpublished results; software can be downloaded at http://www.hkbic.cuhk.edu.hk/software/omblast). RefAligner aligns each molecule map to the reference maps by finding the best-matching region using a dynamic programming algorithm. The match score depends on the distribution of fluorescent labels on the molecule and the *in silico* nicking sites in the region on the reference sequence. OMBlast uses a BLAST-like seed-and-extension approach to aligning molecules to the reference. Each molecule map can be split-mapped to multiple locations in the forward or reversed orientation, allowing for detection of large insertions, deletions, and inversions in the SV detection step.

We integrated the two sets of molecule alignments by taking their consensus as follows. If both alignment methods were able to align a molecule, and their aligned locations were similar (within half the length of the molecule), a consensus was reached, and we included the OMBlast result in our integrated list. On the other hand, if only one method was able to align a molecule, we included this alignment directly regardless of the method. Finally, if both methods were able to align a molecule, but the aligned locations were different (more than half the length of the molecule apart), we would exclude both alignments.

Alignment of de novo assembled genome maps to reference maps: We first aligned the molecules to the genome maps, followed by trimming all molecule alignments by three flanking signals. Then we counted the signal coverage on the assembled genome maps based on the number of molecules aligned. The coverage of the start and the end of the assembled genome maps was usually low, and hence the first and last five signals on the assembled genome maps were ignored. Signals with coverage lower than five supporting alignments were regarded as the low-coverage regions. After splitting the maps at these low-coverage regions, we used OMBlast to align the split fragments with a minimum of 10 signals to reference maps and to detect large insertions, deletions, and inversions using partial-alignment-based strategy as described below, except that the minimum number of fragments supporting the variant call was set to one.

Table 1 Statistics of single-molecule maps and *de novo* consensus maps

	NA12878[a]	NA12891[b]	NA12892[b]
Single-molecule maps			
No. of DNA molecules (k)	994	720	650
Average size (kb)	278	326	328
Maximum size (kb)	2258	2,912	3,255
Total molecule length (Gb)	276	235	213
Estimated average depth of coverage (×)	92×	78×	71×
Consensus maps			
Total consensus map size (Gb)	2.9	3.0	3.0
No. consensus maps	1049	990	995
N50 (Mb)	4.59	4.87	5.00
Longest consensus map size	26.4	25.4	29.0
% aligned to hg38	99%	99%	98%
hg38 genome coverage	96%	96%	96%

[a] Statistics were based on DNA molecules that are >180 kb.
[b] Statistics were based on DNA molecules that are >150 kb.

Identification of structural variants: The integrated list of molecule alignments was then used to identify SVs using a probabilistic method and a partial-alignment-based method. In total, six different types of variations were identified, namely single-site insertions, single-site deletions, segmental insertions, segmental deletions, inversions, and translocations. The first two types involved single nicking sites that were observed on the molecules only but were not present on the reference, or vice versa. These variations could be due to SVs or smaller-scale mutations such as single-nucleotide variants or small insertions and deletions. Since the current study focuses on SVs, below we consider only the latter four types of variants identified by the two methods.

In the probabilistic method, an error model was defined to describe various types of errors possibly contained in the data, including incomplete enzymatic digestion, non-sequence-specific nicks, molecule stretch variation, measurement resolution, and errors. The model parameters were estimated based on the alignment results. For every two adjacent nicking sites on the reference map or that were supported by at least one molecule, the model was used to compare the likelihoods of the null hypothesis of having no SVs between them with four alternative hypotheses, namely having homozygous/heterozygous insertions and having homozygous/heterozygous deletions between the two sites. SVs were then called for cases having a significant *P*-value and alternative-to-null likelihood ratio.

The partial-alignment-based method examined molecules that aligned to multiple genomic regions. SVs were called depending on (1) whether the two aligned regions were on the same chromosome, (2) the distance between the two aligned regions if they were on the same chromosome, and (3) the orientations of the two alignments. Small inversions were also identified by checking if an inverted pattern was observed in the Compact Idiosyncratic Gapped Alignment Report (CIGAR; https://samtools.github.io/hts-specs/SAMv1.pdf) string of an alignment. For example, the CIGAR string of IIMDD (two inserted nicking sites followed by a match followed by

Figure 2 Genome coverage of *de novo* assembled genome maps on hg38. The genome maps of the NA12878 (orange), NA12891 (blue), and NA12892 (red) were aligned to hg38 *in silico* BspQI maps (gray line below NA12878 genome maps). Telomere and centromere locations (green) were based on annotations from the UCSC Table Browser, and N-base gap regions are gray.

two deletions) would be considered a potential inversion since the inversion of the sequence could exactly explain the CIGAR string. Finally, SVs identified from different molecules that were aligned to nearby regions were merged into a single SV.

To minimize the number of false positives, both the probabilistic and partial-alignment-based methods required a minimum number of molecules supporting the variant allele for an SV to be called.

NGS-based validation of BioNano-based SV calls

We attempted to use publicly available NGS data derived from cell lines of the same individuals to validate BioNano-based SV calls. We looked at both the changes in the coverage depth and the changes in the number of unpaired alignments around the SV regions for confirmation. We expected the coverage depth to drop around deletion regions. There might be fluctuation around insertion breakpoints, but the depth was expected to be relatively normal except for duplication events. We also expected an increase in the number of unpaired alignments around SV breakpoints.

Alignment .bam files for the Illumina high-coverage paired-end 100-base data for the trio samples were downloaded from the Genome in a Bottle Consortium (ftp://ftp-trace.ncbi.nih.gov/giab/ftp/data, accessed in August 2013). The alignment was done against hg19. The BioNano-based SV calls were in hg38 coordinates, so they were converted via the batch LiftOver tool (UCSC Genome Browser) into hg19 coordinates.

Calculation of s/(s + p) ratios: From the .bam files, we extracted properly paired alignments using SAMtools (Li *et al.* 2009) with the command "samtools view -f 3." We extracted unpaired (or single-end) alignments using "samtools view -f 4 -F 264" and "samtools view -f 8 -F 260." We divided the genome into 500-bp nonoverlapping bins. For each bin, we counted the number of single-end aligned (denoted as "s") reads and all properly paired aligned (denoted as "p") reads. We then calculated the ratio between single-end aligned reads and all aligned reads (including properly paired reads and single-end aligned reads). Peaks were called for each region. If a peak overlapped with the BioNano SV region, the SV was considered supported.

Calculation of normalized coverage depth: The alignments from the three samples were pooled, and the total number of properly paired reads per bin was determined. GC content (%GC) was calculated for each 500-bp nonoverlapping bin. Ten thousand random bins for each chromosome were randomly sampled for fitting. A Loess fit was applied to model the correlation between %GC and coverage counts. Zero-coverage bins were removed to avoid N-base regions. The normalized counts were obtained by subtracting the per-bin coverage counts from the GC correction minus the median coverage count of the sample. The coverage profiles were segmented using the R/Bioconductor package DNAcopy. The minimum width of a segment was set to be five bins. If a significance

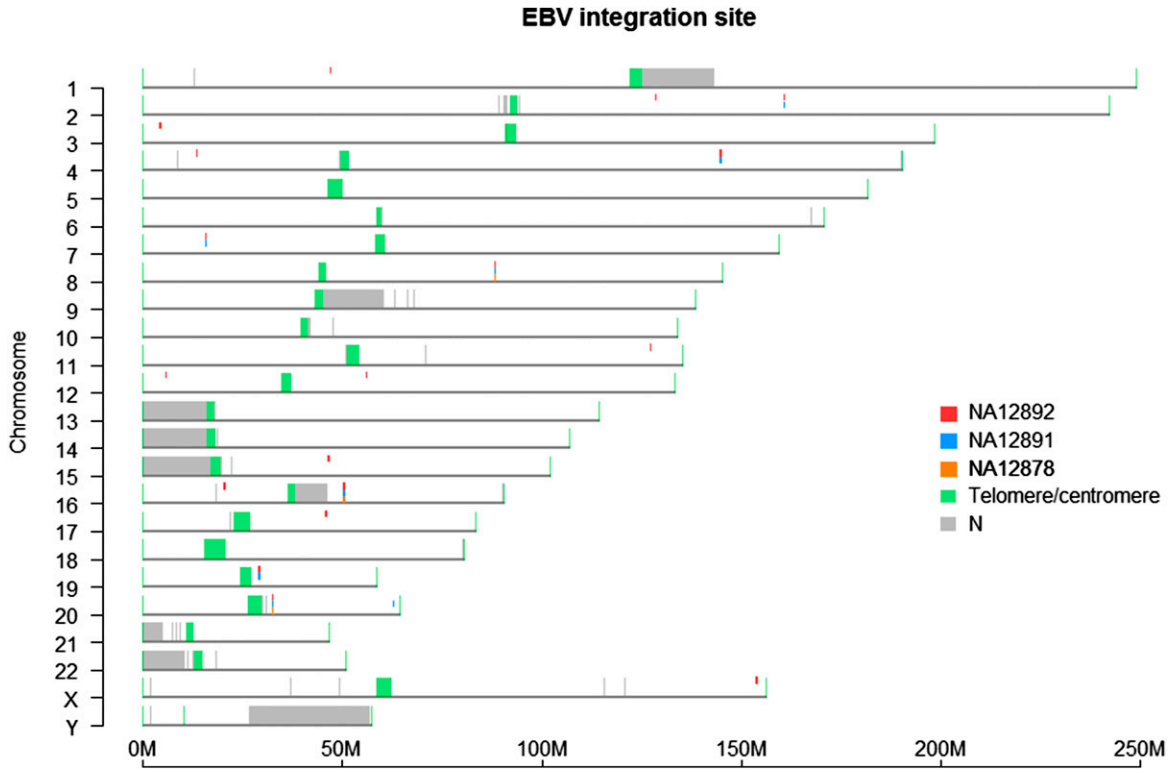

Figure 3 EBV integration sites. Using single-molecule maps that aligned to the EBV genome, we found the potential EBV integration sites by masking the EBV-matching part of the molecules and mapping the rest of the molecules to hg38. Each predicted site was supported by at least 20 molecules. The very different patterns of EBV integration in the trio are most certainly the consequence of the passage of immortalized cell lines in culture and not due to true infection of inheritance from parent to child.

coverage change overlapped with the BioNano SV region, the SV was considered supported.

Gene annotations of structural variants

Homozygous deletions that overlapped with gene exons were identified using ANNOVAR based on RefSeq Gene hg38 coordinates. Among genes located in deleted regions, candidate genes related to disease susceptibility were identified based on the Online Mendelian Inheritance in Man (OMIM) database as well as a literature review. For these candidate genes, deletion boundaries were refined by examining NGS coverage depth and s/(s + p) ratios, as described above. We finalized the list of deletions of genes related to disease susceptibility by verifying that the updated deletion boundaries still overlapped with gene exons.

Results

We applied a genome-mapping strategy that uses long DNA molecules with fluorescent labels marking a specific DNA sequence motif, GCTCTTCN^, for *de novo* genome assembly and structural variation detection. An overview of our genome mapping strategy is outlined in Figure 1.

De novo genome assembly of a CEU (Northern European from Utah) trio

The trio samples (NA12878, NA12891, and NA12892) used in this study came from the CEU collection. We prepared long DNA molecules from the cell lines and labeled them with fluorescent nucleotides (Alexa 546-dUTP), specifically at nicking sites created by nicking endonuclease Nt.BspQI. The labeled DNA molecules were then stained with the DNA intercalating dye YOYO-1 and introduced into nanochannel arrays for imaging. We collected single-molecule data (>150 kb) to a minimum of 71× depth of coverage for each individual (Table 1). The image of each molecule was converted to a single-molecule map based on molecular length and the locations of the fluorescent labels. The single-molecule maps were assembled *de novo* into genome maps. The number of genome maps for each individual ranged from 990 to 1049 (Table 1). Over 98% of genome maps aligned to hg38 and covered at least 96% of hg38 (Table 1 and Figure 2). The remaining 2% of the genome maps correspond to genomic regions substantially different between the trio and the reference genome or with incomplete genomic sequences in the reference (Pendleton *et al.* 2015). The N50 of the three genome assemblies ranged from 4.6 to 5.0 Mb, with the longest map being 29.0 Mb.

In addition to molecules derived from the human genome, we also observed some molecules (0.05–0.40%) aligning to the 170-kb EBV genome. Using molecules partially aligned to EBV, we identified 3 to 18 high-confidence EBV integration sites in these samples (Figure 3). The difference in EBV integration sites between the three samples is most likely due to

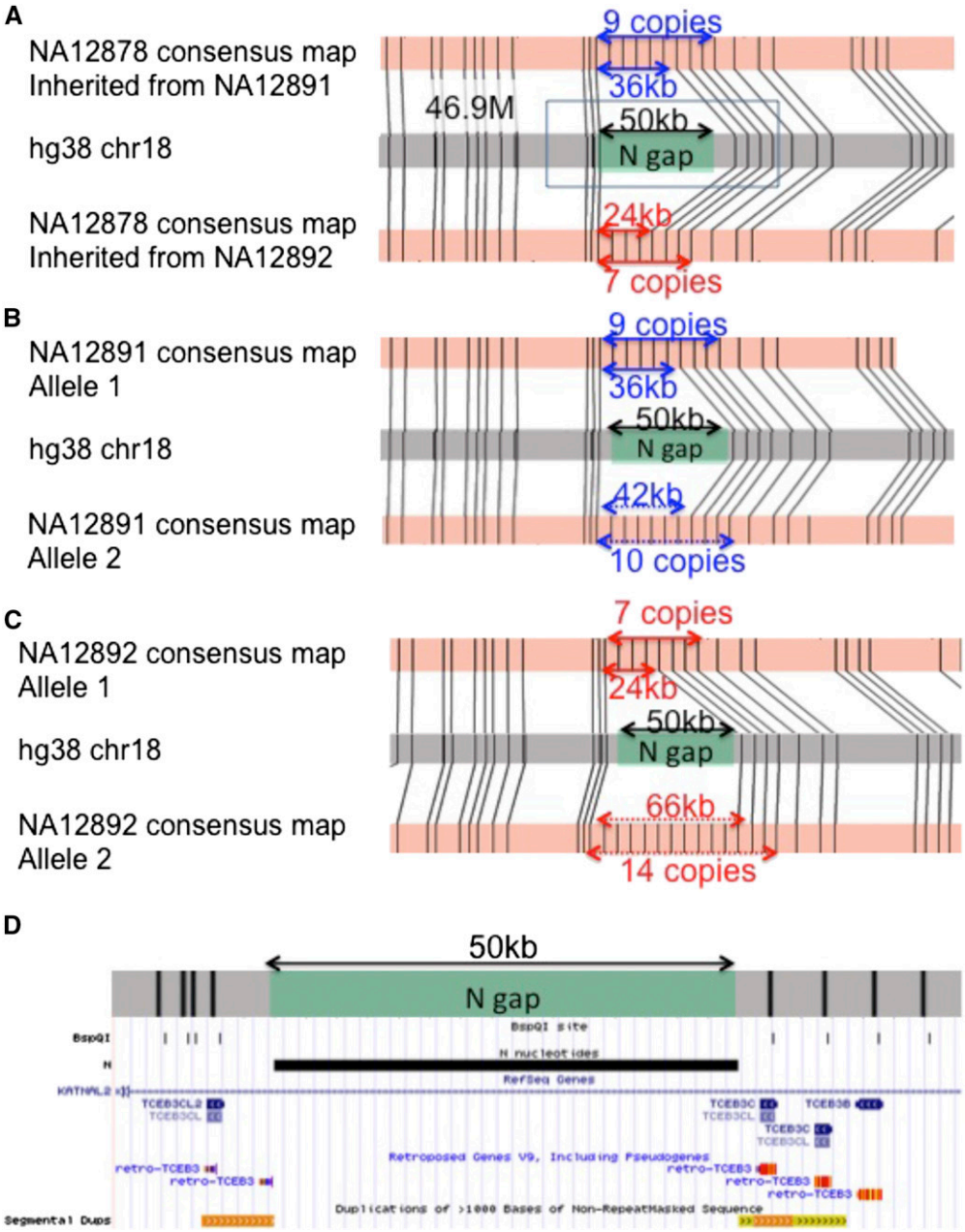

Figure 4 Use of genome maps to size N-base gaps and resolve regions with 6-kb tandem repeats. (A) A 50-kb N-base gap at the chr18:47M region was sized in the trio. NA12878 is heterozygous with 7 and 9 tandem repeats, giving N-base gap sizes of 24 kb (red arrows, inherited from mother, NA12892) and 36 kb (blue arrows, inherited from father, NA12891). (B) NA12891 has 9 and 10 tandem repeats, giving N-base gap sizes of 36 kb (blue arrows) and 42 kb (blue dotted arrows). (C) NA12892 has 7 and 14 tandem repeats, giving N-base gap sizes of 24 kb (red arrows) and 66 kb (red dotted arrows). (D) UCSC genome browser view of the region marked by black box in A. *In silico* BspQI map shown by the BspQI track overlapped with TCEB genes. The variable number of repeats in this region may thus reflect a copy number difference of the TCEB3 family of genes.

a random EBV integration event during passaging of the immortalized cell lines. We also observed that the EBV genome integrated into the lymphoblastoid cell lines was slightly different from the EBV *in silico* map released with the hg38 genome (Supporting Information, Figure S1).

Sizing N-base gaps in hg38

In hg38, there are 267 gaps (>5 kb) that are represented as "Ns" with arbitrary lengths, including 65 located in the subtelomeric and subcentromeric regions (gray in Figure 2). Our *de novo* genome maps spanned across 95 of the 202 N-base gaps outside of known telomeric/centromeric regions, allowing the use of genome maps to estimate actual gap size. As an example, accurate sizing of a 50-kb N-base gap at the hg38 chromosome 18 (47M) region is shown in Figure 4. Genome maps in this region reveal that it represents a copy-number variation where the trio samples contained 7–14 copies of a 6-kb tandem repeat. In contrast to the reference genome, which contains 3 copies of the tandem repeat, NA12878 is heterozygous with 7 and 9 copies, giving N-base gap sizes of 24 and 36 kb, respectively, while NA12891 has 9 and 10 copies (36- and 42-kb gaps), and NA12892 has 7 and 14 copies (24- and 66-kb gaps). In this particular case, the 6-kb tandem repeat resides in the TCEB3 gene locus and is likely to affect the number of copies of the TCEB3 gene (Figure 4D).

Detection of structural variants

Detection of SVs that are >5 kb in size was performed using two complementary approaches, a consensus-based approach and a single-molecule approach. This was followed by manual

inspection of every identified SV to confirm that the SV calls were supported by the presence of single molecules containing the variants. In heterozygotes, the region must be covered with sufficient depth (>50×) and the variant seen in >20% of the molecules to be considered valid. As shown in Table 2, we identified 909 insertions and 661 deletions (>5 kb in size) in these three samples, significantly more than the 59 insertions and 156 deletions previously found (1000 Genomes Project Consortium *et al.* 2010, 2012). We also identified a total of 27 inversions and 44 large copy-number variants in these individuals.

Insertion and deletion

We first examined the single-molecule maps for the loci where 59 insertions and 156 deletions (>5 kb) were previously reported in the 1000 Genomes Consortium pilot and phase 1 data (1000 Genomes Project Consortium *et al.* 2010, 2012). Our maps provided supporting evidence for 39 reported insertions (66%) and 125 reported deletions (80%) (Table 2, "By novelty. . ."). For the rest of the reported insertions and deletions, our data showed that either the reported SVs were not present (11 insertions and 21 deletions) or the SVs present were of a type different from those reported (2 insertions). Fifteen of the reported SVs (7 insertions and 8 deletions) were not detected because our maps did not have sufficient depth to make an SV call. Apart from validating previously reported insertions and deletions, our single-molecule maps and genome maps identified an additional 870 novel insertions and 536 novel deletions (Table 2, "By novelty. . .") with a Mendelian concordance rate of 96% (Table 2, "By Mendelian inheritance"). All these insertions and deletions were carefully verified manually to confirm supporting evidence from single-molecule maps (Table S1 and Table S2). Over 85% of the deletions were validated by next-generation sequence data using read-depth or single-end or paired-end ratio approaches (Table S3).

Among the deletions identified, we found homozygous deletions that affected five genes (*GSTM1*, *LCE3B/C*, *CR1*, *SIGLEC14*) that may affect disease susceptibility (Table 3). After refining deletion boundaries using short-read sequencing data, the *GSTM1*, *LCE3B*, *and LCE3C* genes were completely deleted while the last four exons were deleted from *SIGLEC14* and two exons were deleted from *CR1*. Complete deletions of *GSTM1*, *LCE3B*, *and LCE3C* have previously been reported. *GSTM1* deletion is associated with reduced metabolism of chemical carcinogens and other toxins, leading to increased susceptibility to multiple cancers as well as other disorders such as aplastic anemia (Zhong *et al.* 1993; Trizna *et al.* 1995; Lee *et al.* 2001). Deletion of *LCE3B/C* is a known risk factor for psoriasis (De Cid *et al.* 2009). A null allele of *SIGLEC14* has been associated with increased susceptibility to group B *Streptococcus* (Ali *et al.* 2014), while polymorphisms in *CR1* determine the Knops system blood group and have been associated with resistance to malaria (Cockburn *et al.* 2004).

Table 2 Validated insertions and deletions (>5 kb) detected by single-molecule maps and genome maps

Sample/SV Category	Insertion	Deletion
By samples		
NA12878	769	522
NA12891	743	496
NA12892	748	456
By novelty based on 1000 Genomes Project pilot and phase 1 insertions and deletions >5 kb		
Known	39	125
Novel	870	536
By Mendelian inheritance		
Mendelian	879	631
Non-Mendelian	4	4
No call[a]	26	26
Total	909	661

[a] Unable to generate a call for Mendelian inheritance due to insufficient data to determine an insertion/deletion call to any of the samples.

Inversion

We attempted to verify 51 inversions previously reported in NA12878 by Kidd *et al.* (2008). Of these, our genome mapping data provided single-molecule support for only 16 inversions (31%). An example is found in Figure 5A. Ten of the 16 detected inversions showed partial symmetry in the BspQI nicking pattern, suggesting that these previously reported inversions are palindromes. This hypothesis was verified by generating a dot plot of these 10 hg38 regions (data not shown). For the remaining 35 published inversions, 11 reside in complex regions consisting of multiple types of structural variants. The single-molecule data provide no support for inversions in the remaining 24 regions. Since these inversions were originally identified in the GRCh35 (hg17) reference map, we used the National Center for Biotechnology Information Genome Remapping Service to map the inversions together with 80-kb flanking regions from hg38 to hg17. We observed that 12 of 24 of the regions containing the unverified inversions were significantly different between hg38 and hg17, indicating that half of the discrepancies were due to the use of different reference genome sequence assemblies. In addition, we identified 11 novel inversions in NA12878. In total, we identified 27 inversions in NA12878, including 16 published and 11 novel inversions (Table S4). As some of the inversions reside in complex regions with multiple SVs, long single-molecule maps provide long-range information and direct evidence of complex structural variation (Figure 5B).

Copy-number variation

Sixty-two large copy-number variation (CNV) gain regions associated with NA12878, NA12891, or NA12892 have been reported by Conrad *et al.* (2010), Wang *et al.* (2007), Pinto *et al.* (2007), and Cooper *et al.* (2008). Of the 62 CNV gain

Table 3 Deletions in the trio samples that are associated with disease susceptibility and drug response

Chromosome	Start	Stop	NA12878	NA12891	NA12892	Gene(s)	Gene description	Disease susceptibility
1	109687501	109714725	del/+	del/del	del/+	*GSTM1*	Glutathione S-transferase Mu-1	Reduced ability to metabolize certain chemical carcinogens and toxins, increasing susceptibility to various cancers and to aplastic anemia
1	152570855	152621659	del/+	del/del	+/+	*LCE3B*, *LCE3C*	Late cornified envelope 3B, C	Increased susceptibility to psoriasis
1	207535283	207565105	del/+	del/del	del/+	*CR1*	Complement component receptor 1	Malarial resistance; determinant of Knops system blood group
19	51628937	51649902	del/+	del/del	+/+	*SIGLEC14*	Sialic acid binding Ig-like lectin 14	Increased susceptibility to group B *Streptococcus*

regions, 44 regions map to two or more locations in hg38, indicating that they are known segmental duplications in hg38. Four of these segmental duplications are located in the subtelomeric regions, consistent with previous findings that there are paralogous blocks of subtelomeric repeat elements (Stong *et al.* 2014). The remaining 18 published CNV gain regions cannot be located in our genome maps.

Resolving zygosity of structural variation

With long single-molecule data, zygosity of structural variation can be directly observed when molecules spanning each haplotype are present, and they can be used to build haplotype-resolved genome maps. An example is illustrated in Figure 1E in which a previously reported 9-kb deletion in NA12892 was found to be heterozygous. In addition, a novel 4-kb insertion was found right next to this deletion locus and is out of phase with the 9-kb deletion, allowing us to construct the haplotypes based on these two structural variants.

Discussion

The importance of having long-range information in genome assembly (and, by extension, whole-genome SV detection) has been demonstrated in recent studies using genome maps, long-read sequences, fosmid libraries (Cao *et al.* 2015), or a combination of these for *de novo* assembly of the human genome (Cao *et al.* 2014, 2015; Pendleton *et al.* 2015). In this report, we focus on the use of long single-molecule and genome maps for SV detection without sequencing or cloning. The single-molecule and *de novo* genome maps that we generated for a well-studied CEU trio allowed us to efficiently identify structural variants >5 kb.

The *de novo* genome maps that we produced with single-molecule data alone have assembly N50 of ≥4.6 Mb. Insertions, deletions, and inversions >5 kb were detected and validated with single-molecule maps and/or *de novo* genome maps against the human reference genome and against each other. Overall, we detected and validated seven times more large insertions/deletions than previously found in the 1000 Genome Consortium pilot and phase 1 data (1000 Genomes Project Consortium *et al.* 2010, 2012). A likely explanation of this discrepancy is that it is harder to detect large insertions with NGS paired-end reads with small inserts. Generally, NGS reads help identify insertion breakpoints, but additional assembly is required to identify the inserted sequence. Large insertions are quite obvious with single-molecule and/or *de novo* genome maps. The major advantage of using single-molecule maps is the direct, inference-free supporting evidence for the presence of an SV provided by these maps.

Inversions are usually located at structurally complex regions associated with microdeletions, segmental duplications, and clustered regions of the X chromosome (Kidd *et al.* 2008). Consistent with this observation, we found that at least 22% of the previously reported inversions are embedded in regions with complex structural variants. The majority of the previously reported inversions were not detected by genome mapping partly because inversions found using an older version of the reference genome (hg17) were no longer present in the latest version (hg38). The genome-mapping strategy provides high-level map information to differentiate simple inversion from the more challenging palindromes, especially when the nicking site patterns are easily resolved with long DNA molecules. For SVs larger than the span of single molecules, *de novo* genome maps assembled from single-molecule maps provide an additional level of supporting evidence.

Structural variants are known to be associated with diseases (Weischenfeldt *et al.* 2013). Using our genome-mapping strategy, we identified at least four homozygous deletions that disrupt genes that are known to affect disease susceptibility (Table 3). These genes include *GSTM1*, *LCE3B/C*, *CR1*, and *SIGLEC14*. While we do not have clinical information about these anonymous DNA donors to associate these genotypes with phenotype, genome mapping revealed gene disruptions that may have clinical implications.

With native long DNA molecules without amplification, nonhuman DNA integrated into the genome, such as the EBV genome found in the transformed cell lines studied, can be detected readily. Using single-molecule maps that partially aligned to EBV reference maps and partially to the hg38 reference maps, we determined the EBV integration sites in all three samples. Although EBV integration in these cell lines is only an artifact of the transformation process, genome mapping has potential applications in the study of viral integration in diseases.

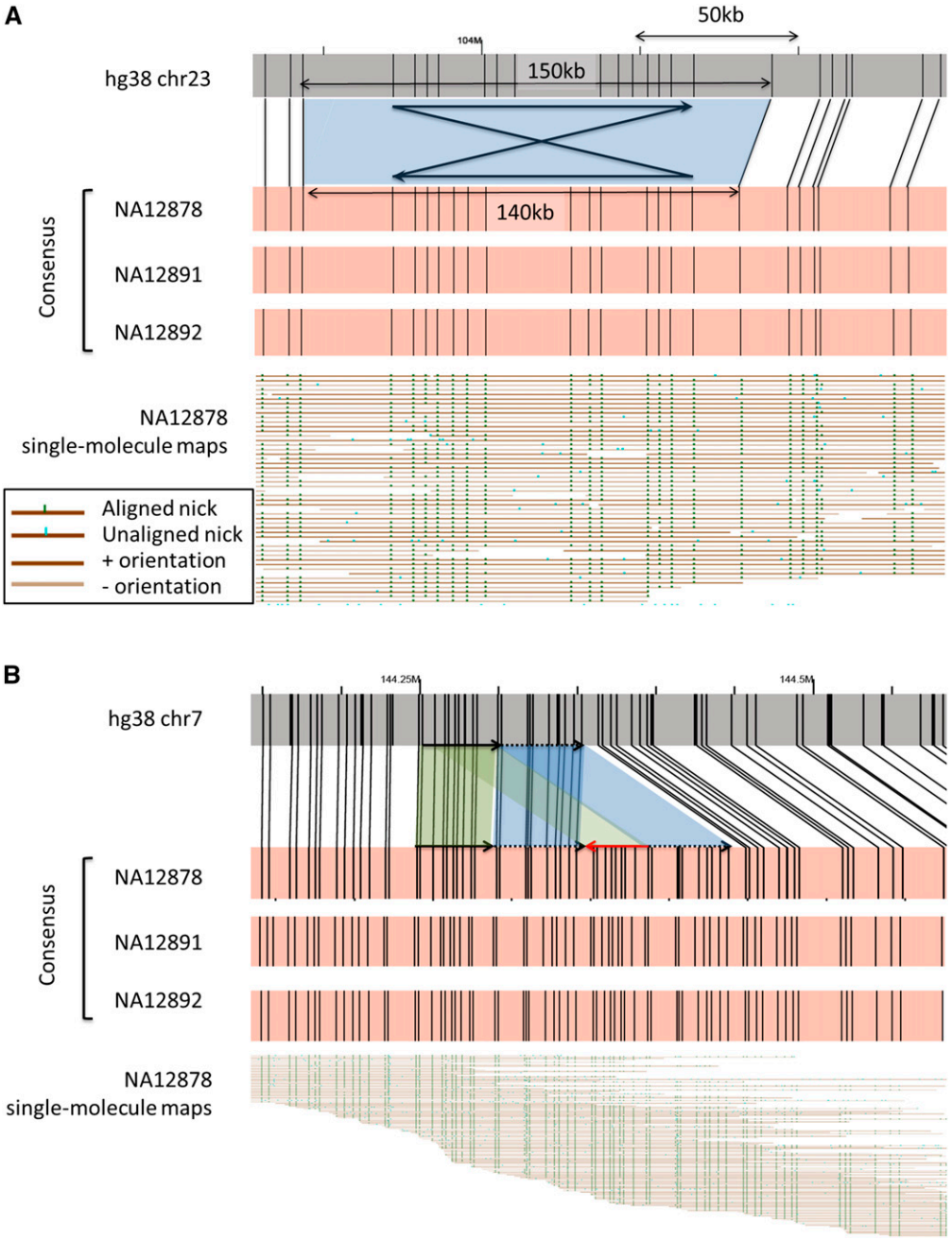

Figure 5 Detection of inversions and complex structural variants. (A) A 50-kb inversion previously reported in NA12878 at the chromosome 23 (104 M) region. A 10-kb deletion (from 150 kb in hg38 to 140 kb in trio) was also detected over the inversion region (blue). This inversion was homozygous in all members in the trio. (B) Detection of complex structural variants. A complex structural variation was detected at the chromosome 7 (144.25 M) region where an inversion was previously reported (Kidd *et al.* 2008). Our genome maps show that this is a structurally complex region: both duplication and inversion events were observed at this locus.

While genome mapping has a number of advantages over current approaches in whole-genome SV detection, there are four major limitations. First, the resolution of the imaging system and uncertainties in DNA measurements keep the method from resolving fluorescent labels that are within 1–2 kb of each other—hence our focus on SVs that are >5 kb in length. With engineering and algorithmic advances, one may be able to improve the mapping resolution. Second, the presence of neighboring nicking sites on opposite strands creates "fragile sites" leading to double-stranded breaks during the nick-labeling process that cannot be bridged by any DNA molecules. For the enzyme used in this study, Nt.BspQI, these fragile sites occur, on average, every 1 Mb in the genome, keeping the genome map N50 to <5 Mb. Bridging the fragile sites requires other data sets, such as long-read sequences or genome maps created by a second nicking enzyme. The third limitation is partly a result of the first two limitations and partly because of the complexity of the human genome. To reduce the number of fragile sites present and keep the fluorescent labels from being too close to each other, we have chosen an enzyme that nicks the human genome at an average interval of 8 kb. This choice has several consequences. Because genome mapping relies solely on the repetitive nicking patterns to detect CNVs, but the labels are ~8 kb apart, only large CNVs containing multiple labels with a unique nicking pattern can be detected when the extra copies are not in tandem of each other. The sparseness of nicking sites makes it hard to pin point the breakpoints of

the SVs detected to less than the 8-kb interval between the outermost pairs of nicking sites marking an SV. For balanced SVs like inversions, reliable detection depends on the number of BspQI nicks and the uniqueness of the nick pattern. Smaller inversions or ones without a unique pattern will not be detected by genome mapping alone. A possible solution is to use two nicking enzymes to create two different maps in separate experiments or to nick-label the same sample with two nicking enzymes to create a doubly labeled map. While these maps can theoretically improve map length and assembly accuracy, added efforts and cost are required with the former approach and the number of fragile sites are increased with the latter approach. Furthermore, the complex regions of the genome will likely pose similar challenges no matter what enzyme is used because of the near identity of large repetitive sequences. The fourth limitation is that genome mapping alone cannot map the extra copies of large CNVs if they are not found in tandem with the original copy. There are three possible scenarios when the extra copy/duplications are not in tandem to the original copy: (1) When the extra copy is too small to have a unique nicking pattern (with nicking frequency of Nt.BspQ1 at 8 kb in the human genome, it takes at least 40–50 kb to have five or more nicks to give a unique pattern), it is seen as an insertion at the new location and nothing more. (2) In some cases, the extra copy has a unique nicking pattern, but it is not identical to the original copy because it has additional nicking sites, is missing some nicking sites, or has additional insertion/deletions. Consequently, a large insertion is seen at the new location but cannot be linked back to the original copy. (3) It is only when a large insertion found elsewhere in the genome with a nicking pattern identical to the original copy that one can determine with some certainty that they are copies of each other. They are previously published segmental duplications, and we have been able to find them in our analysis. When genome maps are combined with long-read sequencing data and short-read sequencing data, even the small CNVs and those with different nicking patterns can be identified because the genome assembly approaches single-base-pair resolution.

A similar approach to genome mapping is optical mapping pioneered by the Schwartz group where long DNA molecules on solid support are restriction-digested *in situ* and ordered restriction maps are produced by sizing the restriction fragments. It has been in numerous genome assembly studies and, more recently, on cancer genomes (Ray *et al.* 2013; Gupta *et al.* 2015). However, fundamental challenges, in terms of sample preparation, data analysis, and information density, with traditional optical mapping remain and have been discussed previously. Alternative methods such as denaturation mapping in nanofluidic channels also show promise but have not been applied to large genomes (Reisner *et al.* 2010).

Although sequence-level data are required to understand the exact arrangement of SV, genome mapping is an efficient way to detect SVs that are >5 kb. Single-molecule maps provide direct evidence and a clear indication of the presence of SV without the need for inference while the *de novo* genome maps allow the detection of SV larger than the span of molecule maps. Combining short-read and long-read technologies, we will finally be able to characterize a full range of genome variation and assemble phased genome sequences that will in turn increase our understanding of the relationship of genetic variation with phenotypes and diseases.

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Author Contributions: Y.Y.Y.L., C.C., C.L., A.R.H., W.S., S.C., and J.S. generated data. A.K.Y.L., T.-P.K., E.T.L., A.C.Y.M. designed and performed structural variant analyses. T.P.K., A.K.Y.L., E.T.L., A.C.Y.M., A.W.C.P., J.J.K.W., C.M.L.L., Z.D., T.A., W.A., X.Z., and H.D. developed analytical tools. E.T.L., A.P., A.C.Y.M., J.-W.L., and A.K.Y.Y. validated structural variants. Y.M. and Z.D. studied and reviewed the biological relevance of structural variants. P.-Y.K., K.Y.Y., T.-F.C., S.-M.Y., M.X., and H.C. supervised the project. A.C.Y.M., E.T.L., Y.M., P.-Y.K., and K.Y.Y. wrote the manuscript.

Literature Cited

1000 Genomes Project Consortium, G. R. Abecasis, D. Altshuler, A. Auton, L. D. Brooks *et al.*, 2010 A map of human genome variation from population-scale sequencing. Nature 467: 1061–1073.

1000 Genomes Project Consortium, G. R. Abecasis, A. Auton, L. D. Brooks, M. A. DePristo *et al.*, 2012 An integrated map of genetic variation from 1,092 human genomes. Nature 491: 56–65.

Ali, S. R., J. J. Fong, A. F. Carlin, T. D. Busch, R. Linden *et al.*, 2014 Siglec-5 and siglec-14 are polymorphic paired receptors that modulate neutrophil and amnion signaling responses to group B streptococcus. J. Exp. Med. 211: 1231–1242.

Burton, J. N., A. Adey, R. P. Patwardhan, R. Qiu, J. O. Kitzman *et al.*, 2013 Chromosome-scale scaffolding of de novo genome assemblies based on chromatin interactions. Nat. Biotechnol. 31: 1119–1125.

Cao, H., A. R. Hastie, D. Cao, E. T. Lam, Y. Sun *et al.*, 2014 Rapid detection of structural variation in a human genome using nanochannel-based genome mapping technology. Gigascience **3:** 34.

Cao, H., H. Wu, R. Luo, S. Huang, Y. Sun *et al.*, 2015 De novo assembly of a haplotype-resolved human genome. Nat. Biotechnol. 33: 617–622.

Cockburn, I. A., M. J. Mackinnon, A. O'Donnell, S. J. Allen, J. M. Moulds *et al.*, 2004 A human complement receptor 1 polymorphism that reduces plasmodium falciparum rosetting confers protection against severe malaria. Proc. Natl. Acad. Sci. USA 101: 272–277.

Conrad, D. F., D. Pinto, R. Redon, L. Feuk, O. Gokcumen *et al.*, 2010 Origins and functional impact of copy number variation in the human genome. Nature 464: 704–712.

Cooper, G. M., T. Zerr, J. M. Kidd, E. E. Eichler, and D. A. Nickerson, 2008 Systematic assessment of copy number variant detection via genome-wide SNP genotyping. Nat. Genet. 40: 1199–1203.

de Cid, R., E. Riveira-Munoz, P. L. Zeeuwen, J. Robarge, W. Liao *et al.*, 2009 Deletion of the late cornified envelope LCE3B and

LCE3C genes as a susceptibility factor for psoriasis. Nat. Genet. 41: 211–215.

El-Metwally, S., T. Hamza, M. Zakaria, and M. Helmy, 2013 Next-generation sequence assembly: four stages of data processing and computational challenges. PLOS Comput. Biol. 9: e1003345.

English, A. C., W. J. Salerno, O. A. Hampton, C. Gonzaga-Jauregui, S. Ambreth *et al.*, 2015 Assessing structural variation in a personal genome: towards a human reference diploid genome. BMC Genomics **16:** 286.

Gupta, A., M. Place, S. Goldstein, D. Sarkar, S. Zhou *et al.*, 2015 Single-molecule analysis reveals widespread structural variation in multiple myeloma. Proc. Natl. Acad. Sci. USA 112: 7689–7694.

Hastie, A. R., L. Dong, A. Smith, J. Finklestein, E. T. Lam *et al.*, 2013 Rapid genome mapping in nanochannel arrays for highly complete and accurate de novo sequence assembly of the complex Aegilops tauschii genome. PLoS One 8: e55864.

Kaplan, N., and J. Dekker, 2013 High-throughput genome scaffolding from in vivo DNA interaction frequency. Nat. Biotechnol. 31: 1143–1147.

Kidd, J. M., G. M. Cooper, W. F. Donahue, H. S. Hayden, N. Sampas *et al.*, 2008 Mapping and sequencing of structural variation from eight human genomes. Nature 453: 56–64.

Lee, K. A., S. H. Kim, H. Y. Woo, Y. J. Hong, and H. C. Cho, 2001 Increased frequencies of glutathione S-transferase (GSTM1 and GSTT1) gene deletions in Korean patients with acquired aplastic anemia. Blood 98: 3483–3485.

Ley, T. J., E. R. Mardis, L. Ding, B. Fulton, M. D. McLellan *et al.*, 2008 DNA sequencing of a cytogenetically normal acute myeloid leukaemia genome. Nature 456: 66–72.

Li, H., B. Handsaker, A. Wysoker, T. Fennell, J. Ruan *et al.*, 2009 The sequence Alignment/Map format and SAMtools. Bioinformatics 25: 2078–2079.

Pendleton, M., R. Sebra, A. W. Pang, A. Ummat, O. Franzen *et al.*, 2015 Assembly and diploid architecture of an individual human genome via single-molecule technologies. Nat. Methods 12: 780–786.

Pinto, D., C. Marshall, L. Feuk, and S. W. Scherer, 2007 Copy-number variation in control population cohorts. Hum. Mol. Genet. 16 Spec No. 2: R168–R173.

Ray, M., S. Goldstein, S. Zhou, K. Potamousis, D. Sarkar *et al.*, 2013 Discovery of structural alterations in solid tumor oligodendroglioma by single molecule analysis. BMC Genomics **14:** 505.

Reisner, W., N. B. Larsen, A. Silahtaroglu, A. Kristensen, N. Tommerup *et al.*, 2010 Single-molecule denaturation mapping of DNA in nanofluidic channels. Proc. Natl. Acad. Sci. USA 107: 13294–13299.

Rice, P., I. Longden, and A. Bleasby, 2000 EMBOSS: The European Molecular Biology open software suite. Trends Genet. 16: 276–277.

Shelton, J. M., M. C. Coleman, N. Herndon, N. Lu, E. T. Lam *et al.*, 2015 Tools and pipelines for BioNano data: molecule assembly pipeline and FASTA super scaffolding tool. BMC Genomics **16:** 734.

Siegel, A. F., G. van den Engh, L. Hood, B. Trask, and J. C. Roach, 2000 Modeling the feasibility of whole genome shotgun sequencing using a pairwise end strategy. Genomics 68: 237–246.

Stong, N., Z. Deng, R. Gupta, S. Hu, S. Paul *et al.*, 2014 Subtelomeric CTCF and cohesin binding site organization using improved subtelomere assemblies and a novel annotation pipeline. Genome Res. 24: 1039–1050.

Trizna, Z., G. L. Clayman, M. R. Spitz, K. L. Briggs, and H. Goepfert, 1995 Glutathione S-transferase genotypes as risk factors for head and neck cancer. Am. J. Surg. 170: 499–501.

Usher, C. L., R. E. Handsaker, T. Esko, M. A. Tuke, M. N. Weedon *et al.*, 2015 Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. Nat. Genet. 47: 921–925.

Wang, K., M. Li, D. Hadley, R. Liu, J. Glessner *et al.*, 2007 PennCNV: an integrated hidden Markov model designed for high-resolution copy number variation detection in whole-genome SNP genotyping data. Genome Res. 17: 1665–1674.

Weischenfeldt, J., O. Symmons, F. Spitz, and J. O. Korbel, 2013 Phenotypic impact of genomic structural variation: insights from and for human disease. Nat. Rev. Genet. 14: 125–138.

Xiao, S., J. Li, F. Ma, L. Fang, S. Xu *et al.*, 2015 Rapid construction of genome map for large yellow croaker (larimichthys crocea) by the whole-genome mapping in BioNano genomics irys system. BMC Genomics **16:** 670.

Zhong, S., A. H. Wyllie, D. Barnes, C. R. Wolf, and N. K. Spurr, 1993 Relationship between the GSTM1 genetic polymorphism and susceptibility to bladder, breast and colon cancer. Carcinogenesis 14: 1821–1824.

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Genome-Wide Structural Variation Detection by Genome Mapping on Nanochannel Arrays

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Supplementary materials

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Figure S1 Mapping singleM molecule maps to the EBV *in silico* map (blue box). NA12878 single-molecule maps are shown as brown horizontal lines. Our data suggest that the EBV genome present in the trio may be slightly different from the one released with hg38. The inconsistencies are highlighted in the boxed region (red box).

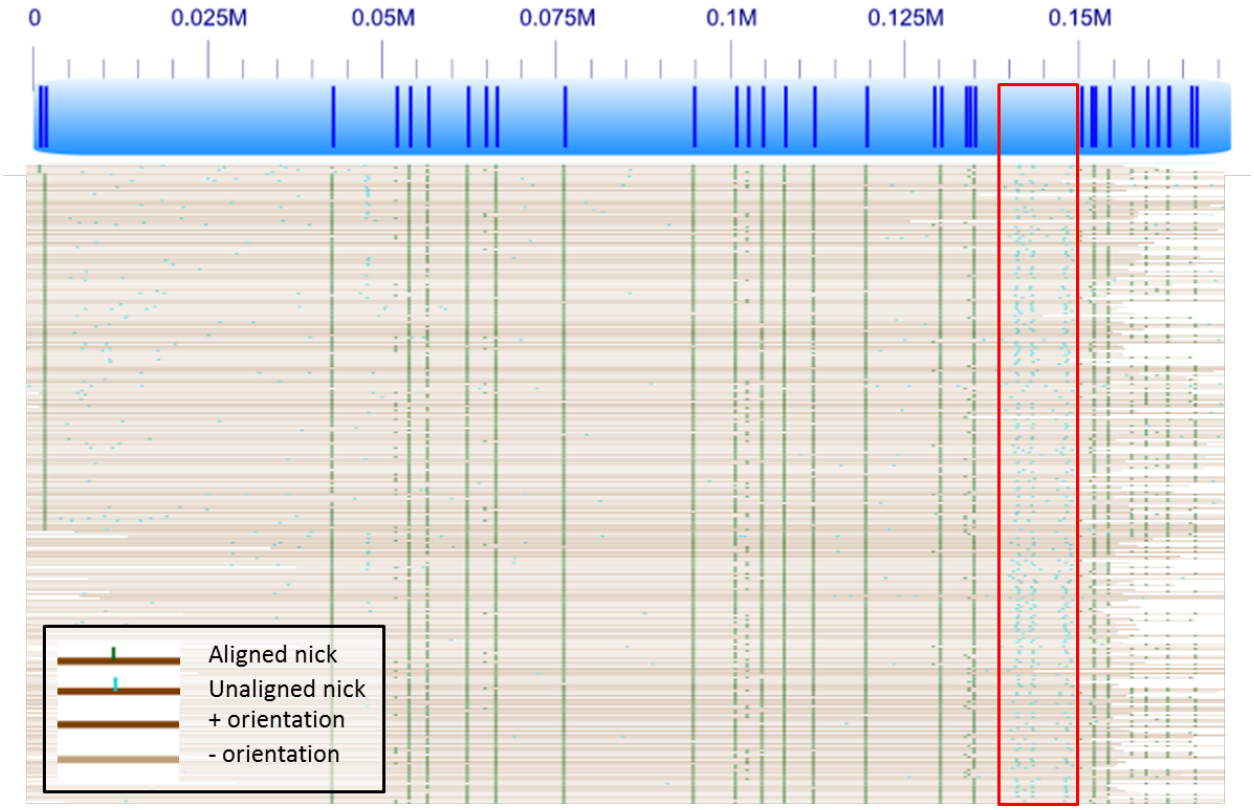

Table S1 Deletion detected in the trio.

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
1	728009	743208	Deletion	del/+	+/+#	del/+#	1		1	1	1	1	1	1	1	1	1
1	13186002	13223978	Deletion	del/+	+/+#	del/del#	1										
1	15812459	15832247	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
1	15827308	15833976	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
1	16564155	16592897	Deletion	del/+	del/+	+/+	1		1	1	1	0	1	0	1	1	1
1	24798267	24863773	Deletion	del/+*	del/del	del/+*	1		1	1	0	1	1	1	1	1	1
1	25255036	25330105	Deletion	del/+#	del/+#	+/+	1		1	1	1	1	0	0	1	1	1
1	34635820	34646375	Deletion	del/+	+/+	del/+*	1	DGV	1	0	1	0	0	0	1	0	1
1	37445605	37479195	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	1	0	1	1
1	39531426	39566732	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
1	56365012	56377267	Deletion	del/+	+/+	del/+	1		1	0	0	1	0	0	1	0	0
1	65558490	65564584	Deletion	del/del	del/+	del/del	1	DGV	1	0	1	1	0	1	1	0	1
1	72300640	72346156	Deletion	del/del	del/del	del/del	1	DGV									
1	75360785	75384497	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
1	78173038	78189682	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
1	80931576	80943725	Deletion	del/del	del/del	del/del	1		1	1	1	1	0	0	1	1	1
1	84052242	84058946	Deletion	del/+	del/del	del/+*	1	DGV									
1	85928603	85951219	Deletion	+/+	del/+	+/+	1		0	0	0	0	0	0	0	0	0
1	89008976	89018424	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
1	108186768	108212805	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
1	108971111	109037022	Deletion	del/+*	del/del*	+/+	1		1	1	0	0	1	0	1	1	0
1	109640831	109652377	Deletion	del/+	NC	del/+#	NC		1	1	1	1	1	0	1	1	1
1	109640831	109711941	Deletion	del/+	NC	del/+#	NC		1	1	1	1	1	0	1	1	1
1	109687501	109714725	Deletion	del/+	NC	del/+#	NC		1	1	1	0	1	0	1	1	1
1	115680250	115690301	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
1	120433799	120457979	Deletion	del/del	del/del	del/del	1										

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
1	146055371	146145645	Deletion	del/+#	del/+#	del/+#	1										
1	146129664	146145645	Deletion	del/+#	del/+	del/+#	1										
1	149055868	149067071	Deletion	del/del*	del/+*	del/+	1										
1	152570855	152621659	Deletion	del/+#	del/del#	+/+	1		1	1	0	0	1	0	1	1	0
1	152758762	152800366	Deletion	del/del	del/+	del/+	1		1	1	1	1	0	0	1	1	1
1	158887634	158909940	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
1	158964562	158997293	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
1	179350407	179372457	Deletion	+/+	del/+#	+/+	1		0	1	0	0	0	0	0	1	0
1	180780632	180786257	Deletion	del/+	del/+	+/+	1	DGV									
1	184842858	184856981	Deletion	del/+	del/+	del/del	1		1	0	1	1	0	1	1	0	1
1	184847145	184856981	Deletion	del/+	del/+	del/del	1		1	0	1	1	0	1	1	0	1
1	187746973	187753401	Deletion	del/del	del/del	del/+	1	DGV									
1	189735377	189814229	Deletion	del/+	+/+	del/+	1	DGV									
1	194477081	194489032	Deletion	del/del	del/+	del/+#	1		1	1	1	1	0	1	1	1	1
1	207359706	207384242	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
1	207523594	207546536	Deletion	+/+	del/+	del/+	1		1	1	0	0	1	0	1	1	0
1	207535283	207565105	Deletion	del/+	del/del	del/+	1		1	1	1	0	1	1	1	1	1
1	207546536	207571864	Deletion	+/+*	del/+	del/+	1		1	1	0	0	1	1	1	1	1
1	209904710	209912632	Deletion	+/+	+/+	del/+	1	DGV									
1	217996859	218027325	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
1	222197929	222204379	Deletion	del/+*	del/+	del/+	1		1	1	1	0	0	0	1	1	1
1	229676786	229685089	Deletion	del/del	del/+	del/+	1	DGV									
1	247687160	247693213	Deletion	del/del	del/del	del/del	1	DGV									
1	247867952	247896148	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
1	247885233	247898339	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
1	248517812	248539532	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
1	248518195	248539532	Deletion	del/del#	del/del	del/del	1		1	0	0	1	1	1	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
2	3674721	3701394	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	1	1	0	1
2	4159347	4177334	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	0	1	0	1
2	4732796	4751638	Deletion	del/del#	del/del	del/del#	1		1	1	1	1	1	1	1	1	1
2	14564045	14569992	Deletion	del/+	del/+	+/+	1	DGV									
2	34470178	34511725	Deletion	del/del	del/del	del/+	1	DGV									
2	34559034	34586785	Deletion	del/+	+/+	del/+	1		1	0	0	1	0	1	1	0	1
2	35736575	35791144	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	1	1	1	1
2	35768573	35791144	Deletion	del/del#	del/+	del/del#	1		1	1	1	1	0	1	1	1	1
2	41011226	41023153	Deletion	+/+	del/+	+/+	1	DGV									
2	41546439	41567598	Deletion	+/+	del/+	+/+	1		1	1	0	0	1	0	1	1	0
2	48553327	48558203	Deletion	del/+	NC	NC	NC		0	0	1	1	1	1	1	1	1
2	49300144	49318399	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
2	49300914	49318399	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
2	52522548	52558137	Deletion	del/del	del/del	del/+*	1	DGV									
2	56425042	56440287	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
2	59396121	59403913	Deletion	del/+	del/+	+/+	1	DGV									
2	64251691	64262298	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
2	76282643	76337471	Deletion	del/del#	del/del#	del/del#	1		1	1	1	1	1	1	1	1	1
2	78848381	78864733	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	1	1	1	1
2	78848381	78865147	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	1	1	1	1
2	88729257	88747641	Deletion	del/del	del/del	del/+	1		1	0	0	1	1	1	1	1	1
2	88862547	88880713	Deletion	+/+#	del/+	+/+	1		1	0	0	0	0	0	1	0	0
2	89016072	89045521	Deletion	+/+	+/+	del/+#	1		0	0	1	1	0	1	1	0	1
2	95885822	95910878	Deletion	+/+	+/+	del/+	1		0	1	0	0	0	0	0	1	0
2	106263101	106269459	Deletion	+/+	del/+	+/+	1	DGV									
2	107655219	107665932	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
2	109065755	109086917	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
2	109929060	109980901	Deletion	del/del#	del/+	del/del	1		1	1	0	1	1	1	1	1	1
2	110390593	110442442	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
2	122715516	122728958	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
2	122727368	122745572	Deletion	del/+	del/+	+/+	1		0	0	0	0	0	0	0	0	0
2	125676257	125684668	Deletion	del/+	+/+	+/+	0		0	0	0	0	0	0	0	0	0
2	125684668	125694984	Deletion	del/+	+/+	+/+	0		1	1	1	0	0	0	1	1	1
2	126910752	126921956	Deletion	del/del	del/del	del/del	1		1	1	1	0	1	1	1	1	1
2	128880845	128889018	Deletion	del/+	del/+	del/+*	1	DGV									
2	129484671	129493838	Deletion	del/+*	del/+	+/+	1	DGV									
2	130853641	130872196	Deletion	del/del	del/+	del/+	1		1	1	0	1	0	0	1	1	0
2	134204720	134220196	Deletion	del/del	del/del	del/+	1		1	1	0	0	0	0	1	1	0
2	138165589	138174944	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0
2	146105051	146119294	Deletion	del/del	del/+*	del/+*	1	DGV									
2	150174624	150181732	Deletion	del/+	+/+	del/+	1	DGV									
2	151570724	151612777	Deletion	+/+	del/+	+/+	1		1	0	1	1	1	0	1	1	1
2	154958360	154986238	Deletion	del/+	del/+	+/+	1		1	1	0	1	0	0	1	1	0
2	159072908	159112035	Deletion	del/+*	del/+*	del/+*	1		1	1	1	1	0	1	1	1	1
2	160585978	160598825	Deletion	+/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
2	169248605	169254652	Deletion	del/+	del/+	del/+	1	DGV									
2	172031326	172054779	Deletion	+/+	del/+	del/+	1		1	1	1	1	1	1	1	1	1
2	172315251	172321376	Deletion	del/+	+/+	del/+	1	DGV									
2	176399231	176412123	Deletion	del/+	+/+	del/del#	1		1	0	1	0	0	1	1	0	1
2	179187833	179214423	Deletion	del/del#	del/del#	NC	NC		1	1	0	0	0	0	1	1	0
2	179197863	179214423	Deletion	del/del#	del/del#	NC	NC		1	1	0	0	0	0	1	1	0
2	179197863	179220326	Deletion	del/del#	del/del#	NC	NC		1	1	0	0	0	0	1	1	0
2	183220793	183226218	Deletion	del/+	del/+	del/+*	1	DGV									
2	190107085	190147829	Deletion	del/+	+/+	del/+*	1		0	0	0	0	1	0	0	1	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
2	193819192	193835350	Deletion	del/del	del/del#	del/del#	1		1	1	1	1	1	1	1	1	1
2	201275689	201284836	Deletion	+/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
2	203014063	203042445	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
2	203311656	203329162	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
2	207484220	207494120	Deletion	del/+	del/+	del/+	1		1	1	1	0	0	0	1	1	1
2	207484220	207494832	Deletion	del/+	del/+	del/+	1		1	1	1	0	0	0	1	1	1
2	209072649	209099554	Deletion	del/+	+/+	del/+#	1		1	0	1	0	0	0	1	0	1
2	216222864	216235751	Deletion	del/del	del/del	del/del	1		1	0	1	1	1	1	1	1	1
2	226300708	226306453	Deletion	del/+	del/+	+/+	1	DGV									
2	231820031	231874626	Deletion	del/del*	del/+	del/+*	1		1	0	0	1	1	1	1	1	1
3	184236	194107	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
3	5491979	5497833	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
3	6608662	6619431	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
3	11062842	11076921	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
3	22050855	22056303	Deletion	del/del	del/del	del/del	1	DGV									
3	32057584	32074884	Deletion	del/+	del/+	del/del	1		1	1	1	0	0	1	1	1	1
3	39716472	39725476	Deletion	+/+	+/+	del/+	1	DGV									
3	65203194	65229073	Deletion	del/+	del/+	+/+	1	DGV									
3	67429890	67452628	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
3	68585619	68591102	Deletion	del/+	del/+	+/+	1	DGV									
3	68684914	68693100	Deletion	del/+	del/del	del/+	1		1	1	1	0	1	0	1	1	1
3	68684914	68701960	Deletion	del/+	del/del	del/+	1		1	1	1	0	1	0	1	1	1
3	77763661	77769678	Deletion	del/+	del/+*	+/+	1	DGV									
3	89445937	89468084	Deletion	del/+#	del/+#	del/+#	1		0	1	0	1	0	1	1	1	1
3	95210806	95277366	Deletion	+/+*	del/+*	+/+	1		0	0	0	0	0	0	0	0	0
3	95746920	95751919	Deletion	del/+	+/+	del/del	1	DGV									
3	99172352	99187205	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	1	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
3	99223901	99230671	Deletion	del/+	del/+*	+/+	1	DGV									
3	111475237	111534413	Deletion	del/+	del/del*	+/+	1		1	1	0	0	1	0	1	1	0
3	112358660	112411443	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
3	130044352	130087902	Deletion	del/del	del/+	del/del	1	DGV									
3	131981517	131995882	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
3	131988842	131995882	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
3	132269401	132276260	Deletion	+/+	+/+	del/+	1	DGV									
3	136302171	136307357	Deletion	del/+*	+/+	del/+*	1	DGV									
3	146667401	146677074	Deletion	del/+*	del/+	del/+*	1	DGV									
3	148562508	148571232	Deletion	del/+	del/del	+/+	1		0	1	1	0	1	0	0	1	1
3	148563886	148571232	Deletion	del/+	del/del	+/+	1		0	1	1	0	1	0	0	1	1
3	159741236	159766013	Deletion	del/+*	+/+	del/+*	1		1	0	1	0	0	0	1	0	1
3	162757356	162892313	Deletion	del/+	del/del*	del/+	1		1	0	1	1	1	1	1	1	1
3	162807722	162835195	Deletion	del/+	+/+	del/+	1		1	0	1	1	1	1	1	1	1
3	162953892	163053464	Deletion	del/+*	del/+*	del/+*	1		1	1	1	1	1	1	1	1	1
3	163037886	163053464	Deletion	del/del#	del/del#	del/del#	1		1	1	1	1	0	1	1	1	1
3	166431251	166497368	Deletion	del/+*	+/+	del/+*	1		1	0	0	0	0	0	1	0	0
3	177574289	177586561	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
3	177663073	177683861	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
3	186862278	186870493	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
3	190534413	190617460	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	1	0	0	1
3	193157540	193167616	Deletion	del/del	del/del*	del/+	1	DGV									
3	195764014	195784456	Deletion	del/+*	del/+*	+/+*	1		1	1	1	1	1	1	1	1	1
3	195782168	195789845	Deletion	del/+*	del/+*	del/+*	1		1	1	1	1	1	1	1	1	1
3	197207174	197212384	Deletion	del/+	del/+	del/+	1	DGV									
3	198116814	198123579	Deletion	del/+*	del/+	+/+	1	DGV									
4	126300	148663	Deletion	del/del	del/del	del/+	1										

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
4	126300	149334	Deletion	del/del	del/del	del/+	1										
4	4121688	4151877	Deletion	del/+	+/+#	del/del	1		1	0	1	1	0	1	1	0	1
4	4121688	4154386	Deletion	del/+	+/+	del/del	1		1	0	1	1	0	1	1	0	1
4	9156793	9212770	Deletion	del/+	del/+*	del/+	1		1	1	1	1	1	1	1	1	1
4	9175954	9212770	Deletion	del/+*	del/+*	+/+#	1		1	0	0	1	1	1	1	1	1
4	10209636	10232943	Deletion	del/+	del/+	del/del	1	DGV									
4	10390521	10400569	Deletion	del/+	del/+	del/+*	1	DGV									
4	19070796	19109297	Deletion	+/+	del/+	del/+	1		0	1	0	1	1	1	1	1	1
4	21137226	21206721	Deletion	del/+	+/+	del/del	1		0	0	0	1	1	1	1	1	1
4	32798326	32808008	Deletion	del/del	del/del	del/del	1		0	0	0	0	0	0	0	0	0
4	34807998	34873863	Deletion	del/del#	del/+#	del/+#	1		0	0	0	1	0	0	1	0	0
4	49083235	49161472	Deletion	del/del	del/+	del/+	1		0	0	0	1	1	1	1	1	1
4	49299967	49309423	Deletion	+/+	+/+	del/+	1		0	1	0	1	1	1	1	1	1
4	59070942	59105035	Deletion	del/+	del/del	del/+	1		1	1	0	1	1	0	1	1	0
4	62803227	62810674	Deletion	+/+	del/+	+/+	1	DGV									
4	63828782	63863763	Deletion	NC	del/del#	NC	NC		0	1	0	0	1	1	0	1	1
4	63828880	63863763	Deletion	NC	del/del#	NC	1		0	1	0	0	1	1	0	1	1
4	70326926	70344040	Deletion	+/+	del/+	+/+	1		1	1	1	0	1	0	1	1	1
4	74711669	74724651	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
4	74712059	74724651	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
4	78347610	78360141	Deletion	del/+	del/del	del/+	1		1	1	1	1	1	1	1	1	1
4	79958832	79979249	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
4	86032438	86061367	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
4	87339366	87378129	Deletion	del/+	del/del	+/+	1		1	1	1	0	1	0	1	1	1
4	90665189	90696345	Deletion	del/del	del/+	del/del	1		1	0	0	1	1	1	1	1	1
4	90665684	90696345	Deletion	del/+	del/+	del/del	1		1	0	0	1	1	1	1	1	1
4	92920064	92943056	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
4	93642799	93647946	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	1	0	0	1
4	93642799	93651369	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	1	0	0	1
4	106678821	106688566	Deletion	+/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
4	107188323	107210908	Deletion	del/del	del/del	del/del	1		1	0	0	1	0	0	1	0	0
4	107301585	107373025	Deletion	del/+#	del/+#	+/+#	1		1	1	0	1	1	1	1	1	1
4	112575295	112599132	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	1	1	1	1
4	114236189	114274932	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
4	114961806	115020175	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
4	115238681	115268206	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	0	1	0	1
4	121361094	121369237	Deletion	+/+	+/+	del/+	1	DGV									
4	138547123	138560615	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
4	138547123	138561277	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
4	138547123	138561940	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
4	144393216	144419281	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
4	144776150	144794017	Deletion	del/+*	del/+	del/+*	1		0	0	0	0	0	0	0	0	0
4	151863481	151875670	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
4	160116472	160173266	Deletion	del/+	del/+	+/+	1		1	1	0	1	0	0	1	1	0
4	160143705	160173266	Deletion	del/+	del/+	+/+	1		1	1	0	1	0	0	1	1	0
4	160957593	160964940	Deletion	del/+*	+/+	del/+*	1	DGV									
4	166755885	166761920	Deletion	del/+	+/+	del/+	1	DGV									
4	172066600	172114307	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	0	1	0	1
4	172506236	172517695	Deletion	del/del	del/del	del/del#	1		1	1	1	1	1	1	1	1	1
4	172506900	172514177	Deletion	del/del	del/del	del/del*	1		1	1	1	1	1	1	1	1	1
4	186163085	186178997	Deletion	del/+	del/del	del/+	1		1	1	1	0	1	0	1	1	1
4	186419907	186438617	Deletion	del/+	del/del	+/+	1		1	1	1	0	1	0	1	1	1
4	186956237	186979867	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	1	1	1	1
4	189136844	189143118	Deletion	del/+*	del/del*	del/+	1	DGV									

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
5	683914	775362	Deletion	+/+	+/+	del/+	1		0	0	0	1	1	1	1	1	1
5	1932495	1955109	Deletion	del/+	del/+	del/del	1		0	0	0	0	0	0	0	0	0
5	12810240	12825737	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
5	13416479	13422741	Deletion	del/+	del/del	del/+	1	DGV									
5	26796589	26801787	Deletion	del/+	del/+	+/+	1	DGV									
5	46270550	46275734	Deletion	del/del*	del/+*	del/del	1	DGV									
5	58033250	58044271	Deletion	del/del	del/del	del/del	1		1	1	1	0	1	1	1	1	1
5	58033250	58046151	Deletion	del/del	del/del	del/del	1		1	1	1	0	1	1	1	1	1
5	58376065	58393231	Deletion	del/+	del/+	del/del	1		1	0	1	0	1	1	1	1	1
5	58385709	58391402	Deletion	del/+	del/+	del/del#	1		1	0	1	0	1	1	1	1	1
5	65702146	65746183	Deletion	del/+*	+/+	del/+*	1		1	0	1	0	0	0	1	0	1
5	69705229	69737913	Deletion	del/+	del/+	del/+	1		0	0	0	0	1	0	0	1	0
5	69732041	69763344	Deletion	del/+	del/+	del/+	1		0	0	0	0	1	1	0	1	1
5	84645803	84662464	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
5	86257578	86268745	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
5	86261910	86268745	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
5	99490459	99508790	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
5	104517644	104524841	Deletion	del/+	del/del	del/+	1	DGV									
5	105096412	105167971	Deletion	del/+*	+/+	del/+	1	DGV									
5	109259373	109265420	Deletion	+/+	del/+	+/+	1	DGV									
5	114989659	114999295	Deletion	+/+	del/+	+/+	1	DGV									
5	118051403	118058188	Deletion	del/+	+/+	del/+	1	DGV									
5	128068847	128075941	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
5	133583273	133589299	Deletion	+/+	+/+	del/+	1	DGV									
5	135776965	135813764	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
5	138464223	138474836	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
5	138464223	138482506	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
5	152074118	152091698	Deletion	+/+	del/+	del/+	1		1	0	0	0	1	1	1	1	1
5	176907647	176969394	Deletion	del/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
5	178679860	178685524	Deletion	del/+	+/+	del/del	1	DGV									
6	19040764	19050061	Deletion	del/+	del/+	+/+	1	DGV									
6	24808175	24830679	Deletion	del/del	del/del	del/del	1		1	0	1	1	1	1	1	1	1
6	26692120	26712245	Deletion	del/+	del/+	del/+	1										
6	26723295	26778386	Deletion	del/+	del/+	del/+	1										
6	29862093	29949658	Deletion	NC	del/del*	+/+	NC		1	1	1	1	1	1	1	1	1
6	31982792	32023037	Deletion	+/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
6	33615197	33620169	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
6	33965350	33976973	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	0	1	1	0
6	48963308	48970896	Deletion	del/+	del/+	NC	NC	DGV									
6	51863330	51894875	Deletion	del/+	+/+	del/+	1		0	0	0	1	0	1	1	0	1
6	54060005	54070030	Deletion	del/del	del/del	del/del	1	DGV									
6	55961159	55981914	Deletion	del/+*	NC	+/+	NC	DGV									
6	57787155	57820737	Deletion	del/+	del/+	+/+	1										
6	58097571	58102586	Deletion	del/+	del/+	+/+	1	DGV									
6	60393044	60427983	Deletion	del/+	+/+	del/del	1										
6	60634275	60662436	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
6	65686134	65704012	Deletion	del/+#	del/+#	+/+#	1		1	1	0	0	0	0	1	1	0
6	66277741	66334977	Deletion	del/+	+/+	del/+	1		1	1	1	1	1	1	1	1	1
6	66294643	66348539	Deletion	del/+	+/+	del/+	1		1	1	1	1	1	1	1	1	1
6	72154148	72163843	Deletion	del/+*	del/+	del/+	1	DGV									
6	73882344	73892723	Deletion	del/del	del/del	del/del	1	DGV									
6	73903479	73910976	Deletion	del/del	del/del*	del/del	1	DGV									
6	76387528	76393575	Deletion	del/+*	del/del	+/+	1	DGV									
6	76404367	76409959	Deletion	del/+	del/del	+/+	1		0	0	0	0	0	0	0	0	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
6	78243367	78319835	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
6	84609971	84618750	Deletion	del/del	del/del	del/+	1		1	1	0	0	1	0	1	1	0
6	85998091	86003534	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	0	1	1	1
6	85998091	86007304	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
6	101477313	101522262	Deletion	del/+	del/+	+/+	1		1	1	1	0	0	0	1	1	1
6	103289589	103315259	Deletion	del/+	del/+	del/+*	1	DGV									
6	111900053	111911562	Deletion	+/+	del/+	del/+	1		0	1	0	0	0	0	0	1	0
6	111907098	111911562	Deletion	+/+	del/+	del/+	1		0	1	0	0	0	0	0	1	0
6	128485660	128516258	Deletion	del/+*	NC	+/+	NC		1	1	0	0	0	0	1	1	0
6	128984943	129007524	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
6	128999919	129007524	Deletion	del/+	del/+	+/+	1		0	0	0	0	1	0	0	1	0
6	128999919	129011992	Deletion	del/+	del/+	+/+	1		0	0	0	0	1	0	0	1	0
6	132034356	132069337	Deletion	del/+	+/+*	del/+	1		1	0	1	0	0	0	1	0	1
6	132366573	132391892	Deletion	del/+	del/+	+/+	1		1	1	0	1	0	0	1	1	0
6	133015998	133031991	Deletion	+/+	del/+	del/+	1		1	0	0	0	1	0	1	1	0
6	133025207	133031991	Deletion	+/+	del/+	del/+	1		1	0	0	0	1	0	1	1	0
6	152702157	152721835	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
6	154791460	154813336	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
7	1816091	1836994	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
7	1816581	1836994	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
7	4579167	4588031	Deletion	del/+	del/del	+/+	1		0	0	0	0	0	0	0	0	0
7	4579167	4597088	Deletion	del/+	+/+	del/+	1		0	0	0	1	0	1	1	0	1
7	4587479	4597088	Deletion	del/+	+/+	del/del	1		0	0	0	1	0	1	1	0	1
7	5842225	5848166	Deletion	+/+	del/+	+/+	1	DGV									
7	12982476	12988925	Deletion	del/+	del/+*	+/+	1	DGV									
7	25003676	25036864	Deletion	del/+	del/del	+/+	1		0	0	0	1	1	1	1	1	1
7	26090887	26117482	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
7	31272095	31281855	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
7	32345314	32354628	Deletion	del/del	del/del	del/del	1		1	1	0	1	1	0	1	1	0
7	32345314	32360942	Deletion	del/del	del/del	del/del	1		1	1	0	1	1	0	1	1	0
7	37425220	37430174	Deletion	+/+	del/+	+/+	1										
7	38330233	38373352	Deletion	del/+#	del/+#	+/+	1		1	1	0	0	0	0	1	1	0
7	49680242	49686301	Deletion	del/del	del/del	del/del	1	DGV									
7	51524941	51531645	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
7	56584786	56590822	Deletion	+/+	del/+	del/+	1	DGV									
7	56694287	56702254	Deletion	del/+	NC	NC	NC	DGV									
7	65479956	65561218	Deletion	del/+	del/+	del/+#	1		1	1	1	1	1	1	1	1	1
7	65481931	65561218	Deletion	del/+#	del/+#	del/+#	1		1	1	1	1	1	1	1	1	1
7	70969523	70979773	Deletion	del/del	del/del	del/+	1		0	0	0	0	0	0	0	0	0
7	73056054	73112195	Deletion	del/+	del/+	del/+	1										
7	74378921	74438779	Deletion	del/del*	del/del*	del/del*	1		1	1	1	1	1	1	1	1	1
7	75270960	75309222	Deletion	del/+	del/+	del/+	1										
7	76526387	76536569	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	1	0	0	1
7	91582827	91591313	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	91585190	91591313	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	93787533	93796180	Deletion	del/del	del/del	del/del	1	DGV									
7	96838169	96860256	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
7	96838782	96860728	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
7	96910658	96928840	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	1	1	0	1
7	97762083	97773481	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	101348139	101365883	Deletion	del/+	del/+	del/+*	1		1	1	0	1	1	1	1	1	1
7	109794954	109824053	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
7	110541908	110548375	Deletion	del/+	del/+	+/+	1	DGV									
7	113100837	113126700	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
7	113770749	113791390	Deletion	del/del	del/del	del/del	1		1	0	1	1	1	0	1	1	1
7	121330045	121389440	Deletion	del/+	+/+	del/+*	1		0	0	0	0	0	0	0	0	0
7	126405832	126411393	Deletion	del/del*	del/+	del/+	1	DGV									
7	127571922	127595147	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	132555329	132585736	Deletion	+/+	del/+	del/+	1		0	0	0	1	0	0	1	0	0
7	142047097	142095820	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	142047097	142527570	Deletion	del/+	+/+	del/+	1										
7	142066836	142100862	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
7	148326780	148338369	Deletion	del/del*	del/del	del/+*	1		1	1	1	0	1	1	1	1	1
7	150033155	150048690	Deletion	del/+	del/+	+/+	1		0	0	0	1	0	1	1	0	1
7	150598752	150624358	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
7	154599973	154613034	Deletion	+/+	del/+	del/+	1		0	1	1	0	0	0	0	1	1
7	156593383	156618640	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	0	1	1	0
7	156594247	156618205	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	0	1	1	0
8	133246	159033	Deletion	del/del	del/del*	del/del*	1		1	0	0	1	1	1	1	1	1
8	644397	649414	Deletion	del/del	del/+	del/+	1	DGV									
8	1390126	1413734	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
8	6988109	7016368	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
8	7259701	7290189	Deletion	del/+	del/del*	+/+	1		0	1	1	1	1	1	1	1	1
8	7493052	7514811	Deletion	del/del	del/del	del/del	1		0	0	0	1	1	1	1	1	1
8	8063298	8135496	Deletion	del/+*	del/+*	del/+*	1		1	1	0	1	1	1	1	1	1
8	25114917	25133477	Deletion	del/+*	del/+	del/+	1	DGV									
8	25189761	25218938	Deletion	del/+	del/+*	del/del*	1		1	1	1	0	0	1	1	1	1
8	32808632	32855019	Deletion	del/del	del/del	del/del*	1		1	1	1	1	1	1	1	1	1
8	39351594	39531914	Deletion	del/+	del/del	del/+	1		1	1	1	1	1	1	1	1	1
8	40022341	40044568	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
8	40916326	40922473	Deletion	del/del	del/del	del/del*	1	DGV									

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
8	42330947	42349908	Deletion	del/del	del/del*	del/+*	1		1	1	1	1	1	1	1	1	1
8	50118531	50125932	Deletion	+/+	del/+*	+/+	1	DGV									
8	62122387	62127833	Deletion	del/+*	+/+	del/+	1	DGV									
8	63794845	63801899	Deletion	del/+	del/+	+/+	1		1	0	0	1	0	0	1	0	0
8	72872702	72880048	Deletion	del/del	del/del	del/+	1		0	0	0	0	0	0	0	0	0
8	72872702	72890493	Deletion	del/del	del/del	del/+	1		0	0	0	1	1	1	1	1	1
8	74449320	74458914	Deletion	del/del	del/+	del/+	1		1	1	1	1	1	1	1	1	1
8	91520149	91528708	Deletion	+/+	+/+	del/+	1	DGV									
8	93060099	93065214	Deletion	del/+*	+/+	del/del	1	DGV									
8	95849365	95870168	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
8	111271497	111284853	Deletion	del/del	del/+	del/+	1		1	1	1	1	0	0	1	1	1
8	113028236	113034538	Deletion	+/+	del/+	+/+	1	DGV									
8	113476274	113486791	Deletion	del/+	+/+	+/+	0		0	0	0	0	0	0	0	0	0
8	119142303	119149203	Deletion	del/+	del/+	+/+	1	DGV									
8	125584369	125592929	Deletion	del/del	del/del	del/del	1		1	0	0	0	1	1	1	1	1
8	126175255	126201835	Deletion	del/+	del/del*	+/+	1		1	1	0	0	0	0	1	1	0
8	128447566	128457481	Deletion	del/del	del/del	del/del	1		1	1	0	1	1	1	1	1	1
8	131263769	131273753	Deletion	del/+	del/+	del/del	1		1	1	1	0	0	1	1	1	1
8	134061472	134078717	Deletion	del/+	del/del	+/+	1		0	0	0	1	1	0	1	1	0
8	136138496	136152294	Deletion	del/del	del/del	del/+	1		1	1	1	0	1	0	1	1	1
8	137781988	137814546	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
8	143616925	143632872	Deletion	del/+*	del/+	del/del*	1	DGV									
9	6675359	6750423	Deletion	+/+	del/+	del/+	1		0	1	1	0	1	0	0	1	1
9	15815308	15821942	Deletion	del/+*	del/+	+/+	1	DGV									
9	17903677	17930194	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
9	22496046	22504893	Deletion	del/del	del/del	del/+	1	DGV									
9	23357184	23378371	Deletion	del/+	+/+	del/+#	1		1	0	1	0	0	1	1	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
9	29092576	29098016	Deletion	del/+	+/+	+/+	1	DGV									
9	39163650	39198266	Deletion	+/+	+/+	del/+*	1		0	0	0	0	0	0	0	0	0
9	40288702	40332564	Deletion	del/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
9	42340492	42349392	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
9	42340492	42352257	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
9	66023482	66043745	Deletion	del/+	del/+	del/+	1										
9	66043745	66087598	Deletion	del/+	del/+	del/+	1										
9	66847020	66922452	Deletion	del/+	del/+	del/+	1										
9	69102762	69144850	Deletion	del/del	del/+	del/del#	1		1	1	1	1	0	1	1	1	1
9	70688910	70721291	Deletion	del/del	del/del	del/del	1		1	1	1	1	0	1	1	1	1
9	70703298	70744387	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
9	87900317	87914632	Deletion	del/+	del/del	+/+	1		0	0	0	0	0	0	0	0	0
9	87914632	87934580	Deletion	del/+	del/del	+/+	1		0	0	0	1	1	0	1	1	0
9	93730936	93752120	Deletion	del/+	del/+	del/+	1		1	1	1	0	0	0	1	1	1
9	98532315	98564389	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
9	101951963	101962410	Deletion	del/+*	+/+	NC	NC	DGV									
9	107253692	107273834	Deletion	+/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
9	107770232	107778739	Deletion	del/del	del/+	del/del	1		1	0	0	1	1	1	1	1	1
9	117752136	117758158	Deletion	+/+	+/+	del/+*	1		0	0	0	0	0	0	0	0	0
9	127883260	127913069	Deletion	+/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
9	130268233	130281206	Deletion	del/+	del/+	del/+	1		1	1	1	1	1	1	1	1	1
9	138178511	138202992	Deletion	del/del*	del/del*	del/del*	1										
10	1225133	1244692	Deletion	+/+	del/+	+/+	1										
10	5245336	5251382	Deletion	del/+	+/+	del/+	1	DGV									
10	5844539	5856047	Deletion	del/+	del/+	del/del	1		1	1	1	0	0	1	1	1	1
10	6369601	6375667	Deletion	del/del	del/del	del/del	1	DGV									
10	13008064	13030785	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
10	20544822	20569170	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
10	20563327	20574468	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
10	30957818	30962847	Deletion	del/+	del/del	+/+	1	DGV									
10	51443916	51454462	Deletion	del/+*	+/+	del/+	1	DGV									
10	53023782	53029070	Deletion	+/+	del/+	+/+	1	DGV									
10	57447179	57484761	Deletion	del/+	del/+	del/del*	1		1	1	1	0	0	1	1	1	1
10	65546737	65555536	Deletion	+/+	+/+	del/+	1	DGV									
10	69521247	69531536	Deletion	del/+	+/+	NC	NC	DGV									
10	70678509	70684772	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0
10	70684772	70692256	Deletion	del/+	del/+	del/+	1		1	1	1	1	1	1	1	1	1
10	76489573	76511126	Deletion	del/del	del/del	del/del	1		1	1	1	1	0	0	1	1	1
10	76489573	76511723	Deletion	del/del	del/del	del/del	1		1	1	1	1	0	0	1	1	1
10	79713147	79751547	Deletion	del/+	+/+	del/del	1		1	0	1	1	0	1	1	0	1
10	89363111	89402491	Deletion	+/+	+/+	del/+*	1		0	0	0	0	0	0	0	0	0
10	92373936	92385203	Deletion	del/del#	del/del#	del/del#	1		1	0	0	1	1	1	1	1	1
10	98920270	98959255	Deletion	del/+	del/+	del/del	1		1	1	1	0	0	1	1	1	1
10	105294984	105309452	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
10	109799247	109820653	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
10	109799631	109820653	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
10	112346399	112361408	Deletion	del/+#	NC	NC	NC		1	1	1	1	0	1	1	1	1
10	115054837	115068327	Deletion	del/+*	del/+*	+/+	1		0	0	0	0	0	0	0	0	0
10	122609462	122631415	Deletion	del/+	del/+	del/+	1		1	1	0	0	1	0	1	1	0
10	130834480	130842338	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
10	131102642	131118191	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
10	131591640	131610858	Deletion	del/del#	NC	del/del#	NC										
11	4244663	4287561	Deletion	del/+	+/+	del/+	1		1	1	0	1	1	1	1	1	1
11	4931191	4972994	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
11	28981446	29004044	Deletion	del/del#	del/del#	del/del#	1		1	1	1	1	1	1	1	1	1
11	31359835	31395981	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
11	47014816	47046528	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	1	1	1	1
11	47040353	47060957	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
11	48570185	48593942	Deletion	del/del#	del/del#	del/del#	1		1	1	1	1	1	1	1	1	1
11	54735177	54759543	Deletion	del/+#	del/del	+/+	1		1	1	0	0	1	0	1	1	0
11	54735177	54786028	Deletion	del/+	del/del	+/+	1		1	1	0	0	1	0	1	1	0
11	61182376	61223526	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
11	61201717	61242352	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
11	82134811	82200938	Deletion	del/+	+/+	del/+	1		1	0	1	1	1	1	1	1	1
11	89878436	89896142	Deletion	+/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
11	93134902	93138176	Deletion	del/del	del/del	del/del	1		0	0	0	1	1	1	1	1	1
11	93134902	93177680	Deletion	del/del#	del/del#	del/del#	1		0	0	0	1	1	1	1	1	1
11	93419293	93443529	Deletion	del/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
11	95434653	95446392	Deletion	del/del	del/+	del/del	1		1	0	1	1	0	1	1	0	1
11	98895523	98936936	Deletion	del/+	del/+	+/+	1	DGV									
11	101682912	101691298	Deletion	del/del	del/del	del/+	1		0	0	0	0	0	0	0	0	0
11	101695056	101704527	Deletion	del/del	del/del	del/+	1	DGV									
11	104394241	104409672	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
11	106913661	106941990	Deletion	+/+	del/+*	+/+	1		0	1	0	0	0	0	0	1	0
11	124900690	124911680	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
11	134732084	134737767	Deletion	del/+	+/+	del/+	1	DGV									
12	183048	188832	Deletion	del/+	del/+	+/+	1	DGV									
12	268330	275668	Deletion	del/+	del/+	+/+	1	DGV									
12	9478805	9587527	Deletion	del/del	del/+	del/del	1		1	1	1	1	1	1	1	1	1
12	9478805	9587940	Deletion	del/del	del/+	del/del	1		1	1	1	1	1	1	1	1	1
12	10394564	10461803	Deletion	+/+	del/+	del/+	1		0	1	1	1	1	1	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
12	10461803	10477070	Deletion	+/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
12	10465047	10483347	Deletion	+/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
12	22415799	22431905	Deletion	del/del	del/del	del/del	1		1	1	1	1	0	0	1	1	1
12	30084112	30091459	Deletion	+/+	del/+	+/+	1	DGV									
12	30320072	30330376	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
12	33230158	33255543	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	1	0	0	1
12	37586609	37629964	Deletion	del/del#	del/del#	del/+	1		1	1	1	1	1	0	1	1	1
12	37598258	37629964	Deletion	del/del#	del/del#	del/+#	1		1	1	1	1	1	0	1	1	1
12	45509370	45516156	Deletion	del/+	del/+	+/+	1	DGV									
12	58325913	58339245	Deletion	+/+	del/+	+/+	1		0	1	0	0	1	0	0	1	0
12	59542013	59559078	Deletion	+/+	del/+	+/+	1	DGV									
12	60112405	60140052	Deletion	+/+	del/+	del/+	1		0	1	1	0	0	0	0	1	1
12	66130140	66161915	Deletion	+/+	del/+	+/+	1		0	1	0	1	1	1	1	1	1
12	70190654	70223110	Deletion	del/+	del/del	del/+	1		0	0	0	1	1	0	1	1	0
12	84190376	84225380	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
12	87741849	87759508	Deletion	+/+	+/+	del/+	1		0	0	0	1	0	0	1	0	0
12	90084661	90098202	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
12	98546999	98581179	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
12	99399667	99408992	Deletion	del/+	+/+	del/+	1	DGV									
12	101707398	101715813	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
12	117184123	117208237	Deletion	+/+	+/+	del/+*	1		0	0	1	0	0	0	0	0	1
13	18734854	18812648	Deletion	+/+	del/+#	del/+	1										
13	23055560	23061474	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
13	23055560	23073364	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
13	29641686	29647707	Deletion	del/+	del/+	del/+	1	DGV									
13	31958507	31964664	Deletion	+/+	+/+	del/+*	1	DGV									
13	48951298	48975998	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
13	50474543	50507054	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	0	1	1	1
13	50474543	50507136	Deletion	del/del	del/+	del/del	1		1	1	1	1	0	0	1	1	1
13	66597405	66615647	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
13	71890045	71913299	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
13	72231843	72241374	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
13	72262197	72277798	Deletion	del/+	del/+	del/del	1		1	1	1	1	0	1	1	1	1
13	82591799	82598070	Deletion	del/+	del/+*	+/+	1	DGV									
13	97876445	97882203	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
13	98601618	98623086	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
13	109903594	109946236	Deletion	+/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
13	112275337	112327618	Deletion	+/+	del/+	del/+	1										
13	113979265	113990347	Deletion	del/del	del/del	del/del	1										
14	20055673	20089143	Deletion	del/+	del/+	del/+	1		1	1	1	0	0	0	1	1	1
14	22418704	22452648	Deletion	del/+*	NC	NC	NC		0	0	0	0	0	0	0	0	0
14	22418704	22446094	Deletion	del/+	NC	NC	NC		0	0	0	0	0	0	0	0	0
14	22633805	22653250	Deletion	del/+	del/+	del/+	1		0	1	1	1	1	1	1	1	1
14	23971408	24016173	Deletion	del/+	del/+	del/+	1		1	1	1	1	0	0	1	1	1
14	23978901	24016173	Deletion	del/+	+/+	del/+	1		1	1	1	1	0	0	1	1	1
14	24000821	24016173	Deletion	del/del*	del/del	del/+	1		1	1	1	1	0	0	1	1	1
14	40140611	40148468	Deletion	del/del	del/+	del/del*	1	DGV									
14	48992365	49032991	Deletion	del/+	del/+	del/del	1										
14	54236304	54248913	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
14	56438903	56448205	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	0	1	0	1
14	73523334	73569867	Deletion	del/+	del/+*	del/+	1		1	1	1	0	0	0	1	1	1
14	73773091	73780233	Deletion	del/+	del/+	+/+	1	DGV									
14	79639945	79648706	Deletion	del/+*	del/+	+/+	1	DGV									
14	82023876	82045416	Deletion	del/del*	del/del	del/del	1		1	1	1	0	0	1	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
14	100930844	100991898	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0
14	105096697	105117755	Deletion	del/del	del/+	del/+	1		1	0	0	1	1	1	1	1	1
14	105848740	105917899	Deletion	del/+*	del/+*	NC	NC		1	1	1	1	1	1	1	1	1
14	105866208	105917899	Deletion	del/del*	del/del*	del/del*	1		1	1	1	1	1	1	1	1	1
14	106322895	106369198	Deletion	del/del	del/+	del/del#	1										
14	106652742	106745274	Deletion	del/del	del/+	del/del	1										
15	20361185	20384863	Deletion	del/+	+/+	del/+	1		1	1	1	0	0	0	1	1	1
15	20362630	20384863	Deletion	del/+	+/+	del/+	1		1	1	1	0	0	0	1	1	1
15	23424141	23432978	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
15	28524365	28701498	Deletion	+/+	del/+#	NC	NC		0	1	0	1	1	1	1	1	1
15	54898611	54930560	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
15	54923312	54930560	Deletion	del/del	del/del	del/del	1		1	0	0	1	1	1	1	1	1
15	65523185	65530009	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
15	71402947	71417711	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
15	71413168	71428889	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
15	72088819	72130171	Deletion	del/del	del/+*	del/+	1		1	1	1	1	1	1	1	1	1
15	76591304	76605151	Deletion	del/del	del/del*	del/del	1	DGV									
15	82855999	82880847	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
15	82871030	82880847	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
15	82880847	82903291	Deletion	del/del*	del/del	del/+	1		1	1	0	1	1	1	1	1	1
15	100554196	100565347	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
15	101747832	101769186	Deletion	del/+	+/+	del/+	1		0	1	1	0	1	0	0	1	1
16	14690717	14733119	Deletion	+/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
16	16301630	16367993	Deletion	del/+*	del/del*	NC	NC		0	0	0	1	1	1	1	1	1
16	16830166	16859298	Deletion	del/+	del/+	del/del	1		0	0	0	0	0	1	0	0	1
16	19934073	19958270	Deletion	del/+	+/+	del/+	1	DGV									
16	20451873	20470502	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
16	20451873	20471954	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0
16	25316358	25340183	Deletion	del/+	del/+	+/+	1		0	0	0	0	0	0	0	0	0
16	28676104	28710439	Deletion	del/+	+/+	del/del*	1		0	0	0	1	1	1	1	1	1
16	32464010	32595996	Deletion	+/+	+/+	del/+#	1		1	1	1	0	1	1	1	1	1
16	58578898	58616978	Deletion	+/+	+/+	del/+	1		0	0	0	0	0	0	0	0	0
16	62510429	62516764	Deletion	del/+*	del/+	+/+	1	DGV									
16	85399802	85432984	Deletion	+/+	+/+	del/+	1		1	1	1	1	0	1	1	1	1
16	87240807	87255418	Deletion	NC	del/del#	del/+#	NC		0	0	0	0	0	0	0	0	0
17	5682995	5695775	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
17	12432762	12457176	Deletion	del/del	del/+	del/+	1		1	0	1	1	1	1	1	1	1
17	18389459	18491238	Deletion	del/+	del/+	del/+	1		0	0	0	1	1	1	1	1	1
17	18448724	18509141	Deletion	del/+	del/+#	del/+	1		0	0	0	1	1	1	1	1	1
17	27207151	27224681	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	0	1	1	0
17	28449903	28460761	Deletion	del/+	del/del	del/+	1		1	1	1	1	1	1	1	1	1
17	41241555	41282536	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
17	45722427	45753553	Deletion	+/+	del/+*	del/+*	1		0	0	0	0	0	0	0	0	0
17	45775032	45807301	Deletion	NC	del/+*	del/+*	1		0	0	0	0	0	0	0	0	0
17	46274712	46512106	Deletion	del/+	del/+	+/+	1		1	1	0	1	1	1	1	1	1
17	53523111	53588399	Deletion	+/+#	+/+#	del/+*	1		0	1	0	0	0	0	0	1	0
17	53759882	53791375	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
17	53759882	53792891	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
17	56082816	56095596	Deletion	+/+	del/+	+/+	1	DGV									
17	81336003	81355216	Deletion	+/+	+/+	del/+	1		0	0	0	1	0	0	1	0	0
18	14266389	14306411	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
18	32915724	32921298	Deletion	del/+	del/+	+/+	1	DGV									
18	40679928	40686785	Deletion	del/+	del/+	+/+	1	DGV									
18	41281978	41302265	Deletion	del/+	del/+	del/del#	1		1	1	1	0	0	0	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
18	50169815	50179945	Deletion	del/del*	del/+	del/+	1		1	1	1	0	0	0	1	1	1
18	50170383	50173609	Deletion	del/del	del/+	del/+	1		1	1	1	0	0	0	1	1	1
18	54418967	54443192	Deletion	del/del	del/+	del/del	1		1	1	1	1	1	1	1	1	1
18	66056109	66065306	Deletion	del/+	+/+	del/+	1	DGV									
18	70715958	70738275	Deletion	del/+	del/del	del/+	1		1	1	1	0	1	0	1	1	1
19	8259347	8274133	Deletion	del/del	del/+*	del/+	1		1	1	1	0	0	0	1	1	1
19	8274133	8288996	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
19	8280818	8295656	Deletion	del/del	del/+#	del/+#	1		1	1	1	1	1	1	1	1	1
19	8761281	8780482	Deletion	del/+	NC	del/+*	1		1	1	1	0	0	0	1	1	1
19	12581279	12621231	Deletion	del/del*	del/del	del/del	1		1	1	1	1	1	1	1	1	1
19	14905735	14935862	Deletion	del/+	+/+	del/+	1		1	1	1	0	0	0	1	1	1
19	19723634	19728853	Deletion	+/+	del/+	del/+*	1	DGV									
19	29458300	29479248	Deletion	del/+	del/+	+/+	1		0	0	0	0	0	0	0	0	0
19	29458300	29479723	Deletion	del/+	del/+	+/+	1		0	0	0	0	0	0	0	0	0
19	29887009	29903290	Deletion	del/del	del/del	del/del	1		0	0	0	1	1	1	1	1	1
19	35169042	35175489	Deletion	del/+	NC	NC	NC	DGV									
19	37846466	37855441	Deletion	+/+	del/+	+/+	1		1	1	0	0	1	0	1	1	0
19	40101872	40147822	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	0	1	1	0
19	46119477	46125055	Deletion	del/+	del/+	+/+	1	DGV									
19	50574632	50580035	Deletion	del/+	+/+	del/+	1	DGV									
19	51628937	51649902	Deletion	del/+	del/del	+/+	1		1	1	0	1	1	1	1	1	1
19	51628937	51677687	Deletion	del/+	del/del	+/+	1										
19	53182889	53189753	Deletion	del/del	del/del	del/del	1		0	0	0	1	1	1	1	1	1
19	53182889	53190308	Deletion	del/del	del/del	del/del	1		0	0	0	1	1	1	1	1	1
19	54040866	54065257	Deletion	+/+	del/+	del/+	1										
19	56958975	56969060	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
19	56958975	56980682	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
20	1577251	1610985	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
20	1577251	1618537	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
20	2816932	2826033	Deletion	del/+	del/+	+/+	1		1	1	1	1	1	1	1	1	1
20	21277768	21321823	Deletion	+/+	del/+*	del/+*	1		0	1	1	0	0	0	0	1	1
20	25784644	25846589	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
20	25793143	25857438	Deletion	del/+	del/+	+/+	1		0	0	0	1	1	1	1	1	1
20	32933135	32950211	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
20	34218015	34235115	Deletion	del/del*	del/del*	del/del*	1		1	0	0	1	1	1	1	1	1
20	45561127	45579660	Deletion	+/+	del/+	+/+	1		0	1	0	0	0	0	0	1	0
20	54030525	54041463	Deletion	del/+	del/+	+/+	1	DGV									
20	55857199	55872408	Deletion	del/+	del/del	del/+	1		0	1	1	0	1	0	0	1	1
20	55857199	55873125	Deletion	del/+	del/del	del/+	1		0	1	1	0	1	0	0	1	1
20	55857199	55873842	Deletion	del/+	del/del	del/+	1		0	1	1	0	1	0	0	1	1
20	61941030	61948558	Deletion	del/+	del/del	del/+	1		1	1	1	1	1	1	1	1	1
21	8357265	8389456	Deletion	del/+	del/+	del/+	1										
21	19195801	19201921	Deletion	+/+	del/+	+/+	1	DGV									
21	41928122	41940607	Deletion	del/+	del/+	+/+	1		1	1	0	0	0	0	1	1	0
21	43535013	43557020	Deletion	del/+	del/+	del/+	1		1	1	1	1	0	0	1	1	1
22	16065108	16087804	Deletion	+/+	del/+	+/+	1										
22	17289255	17299102	Deletion	del/+	del/del	del/del	1	DGV									
22	17622947	17664459	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
22	17660451	17675840	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
22	20592040	20603836	Deletion	del/+	+/+	del/del	1		1	0	1	0	0	1	1	0	1
22	21468369	21499418	Deletion	del/del	del/del	del/del*	1		0	0	0	1	1	1	1	1	1
22	21468369	21514909	Deletion	del/del	del/del	del/del*	1		0	0	0	1	1	1	1	1	1
22	22853641	22902064	Deletion	del/+	+/+	+/+	0		1	1	0	1	0	0	1	1	0
22	23849945	23864046	Deletion	del/+	del/+	del/del	1		0	0	0	1	1	1	1	1	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
22	31369248	31388431	Deletion	del/+	+/+	del/+*	1		1	0	1	0	0	0	1	0	1
22	36743886	36752828	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
22	38895305	38911753	Deletion	+/+	del/+	del/+	1		0	1	1	0	0	0	0	1	1
23	1634164	1649871	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
23	11694053	11738452	Deletion	del/del	del/del#	del/+	1		1	1	0	1	1	1	1	1	1
23	11929187	11943497	Deletion	+/+	+/+	del/+	1		1	1	1	0	1	1	1	1	1
23	19332741	19342678	Deletion	+/+	del/+#	+/+	1		0	0	0	0	0	0	0	0	0
23	44445450	44468303	Deletion	del/del	del/del*	del/del	1		0	1	0	0	1	0	0	1	0
23	56764920	56776327	Deletion	del/+	+/+	del/+	1		0	1	1	1	1	1	1	1	1
23	56770925	56776327	Deletion	del/+	+/+	del/+	1		0	1	1	1	1	1	1	1	1
23	67903987	67910456	Deletion	del/+	del/del*	+/+	1	DGV									
23	70510563	70519599	Deletion	+/+	+/+	del/+	1		0	0	1	0	0	0	0	0	1
23	70913143	70942444	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
23	79641401	79676548	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
23	81817938	81842687	Deletion	del/+#	del/del#	del/+#	1		1	0	1	1	1	1	1	1	1
23	81836512	81851844	Deletion	del/del	del/del	del/del	1		1	0	1	1	1	1	1	1	1
23	82720687	82734609	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
23	86336537	86353862	Deletion	del/+	del/del	del/+	1		1	1	0	0	0	0	1	1	0
23	86349337	86353862	Deletion	del/+	del/del	del/+	1		1	1	1	0	0	0	1	1	1
23	93541297	93546505	Deletion	+/+	+/+	del/+*	1	DGV									
23	96041289	96072340	Deletion	del/del	del/del	del/del	1		1	0	1	1	1	1	1	1	1
23	103869677	103901604	Deletion	del/del	del/del	del/+	1		1	1	1	1	1	0	1	1	1
23	103901219	103906257	Deletion	del/del	del/del	del/+	1		0	0	0	0	0	0	0	0	0
23	103986456	104000981	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
23	103986456	104004702	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
23	104066827	104091597	Deletion	del/del*	del/del	de/del*	1										
23	109106836	109115226	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	1	1	0	1

chr	start	stop	sv	NA12878	NA12891	NA12892	Men-delian	Publish-ed in DGV	cnp			spratio			cnp OR spratio		
									878	891	892	878	891	892	878	891	892
23	109106836	109115611	Deletion	del/+	+/+	del/+	1		1	0	1	1	0	1	1	0	1
23	115912522	115950771	Deletion	del/+	del/+	del/+	1		0	0	0	0	0	0	0	0	0
23	127442914	127470087	Deletion	del/+	+/+	del/+	1		1	0	1	0	0	0	1	0	1
23	135719353	135884924	Deletion	del1/del2	del1/del1	del1/del2	1										
23	135798571	135833113	Deletion	del/+	del/del	del/+	1										
23	135815845	135892493	Deletion	del/+	+/+	del/+	1										
23	141405496	141432007	Deletion	del/+	del/del#	del/+	1										
23	141425707	141432007	Deletion	del/del	del/del*	del/del	1										
23	143634330	143673900	Deletion	del/del	del/del	del/del	1										
23	144296306	144331569	Deletion	del/+	del/del	del/+	1		1	0	0	0	1	0	1	1	0
23	155544015	155585219	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
23	155569890	155585219	Deletion	del/del	del/del	del/del	1		1	1	1	1	1	1	1	1	1
24	6246224	6272486	Deletion	NC	del/+	NC	NC		0	1	0	1	1	1	1	1	1
24	6628315	6695753	Deletion	NC	del/del*	NC	NC		0	0	0	1	1	1	1	1	1

Table S2 Insertions detected in the trio.

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
1	137901	142869	Insertion	ins/+#	ins/ins#	+/+	1	
1	865008	911412	Insertion	ins/+	ins/ins	ins/+	1	
1	1264345	1295487	Insertion	+/+	+/+	ins/+	1	
1	1360396	1432511	Insertion	ins/ins	ins/ins	ins/ins	1	
1	2322669	2338777	Insertion	ins/ins	ins/ins	ins/ins	1	
1	3458139	3502328	Insertion	ins/ins	ins/ins	ins/ins	1	
1	3486085	3502328	Insertion	ins/ins	ins/ins	ins/ins	1	
1	5383496	5388051	Insertion	ins/+	ins/+	ins/+	1	
1	5383496	5388670	Insertion	ins/+	ins/+	ins/+	1	
1	5383496	5389289	Insertion	ins/+	ins/+	ins/+	1	
1	5383496	5405710	Insertion	ins/+	ins/+	ins/+	1	
1	9615159	9628763	Insertion	ins/ins	ins/ins	ins/ins	1	
1	9628763	9628763	Insertion	ins/ins	ins/ins	ins/ins	1	
1	10969439	10991681	Insertion	ins/ins	+/+	ins/ins	1	
1	10973487	10991681	Insertion	ins/+	+/+	ins/+	1	
1	16564155	16570618	Insertion	ins/+	+/+	ins/+	1	
1	16564155	16592897	Insertion	ins/+	ins/+	ins/+	1	
1	16564155	16606971	Insertion	ins/+	ins/+	ins/+	1	
1	16737593	16740877	Insertion	ins/ins#	ins/+#	ins/ins#	1	
1	18039043	18063412	Insertion	ins/+	ins/+	ins/+	1	
1	18073980	18080955	Insertion	ins/+	ins/+	ins/+	1	
1	19061218	19084845	Insertion	ins/ins	ins/ins	ins/ins	1	
1	23273395	23295959	Insertion	ins/ins	ins/ins*	ins/ins	1	
1	28335803	28356006	Insertion	ins/+	+/+	ins/+	1	
1	28356006	28374907	Insertion	ins/+	+/+	ins/+	1	
1	30400027	30411475	Insertion	ins/+	+/+	ins/+	1	
1	30400027	30412448	Insertion	ins/+	+/+	ins/ins*	1	
1	31429636	31460799	Insertion	ins/+	ins/+	ins/+	1	
1	31429636	31460804	Insertion	ins/+	ins/+	ins/+	1	
1	41537543	41546389	Insertion	ins/+	ins/ins	+/+	1	
1	45292399	45300165	Insertion	+/+	ins/+	+/+	1	
1	45692284	45714139	Insertion	ins/ins*	ins/ins	ins/ins	1	
1	48959114	48962403	Insertion	ins/+*	ins/+	+/+	1	
1	48963035	48968480	Insertion	ins/+	ins/+	+/+	1	DGV
1	51903590	51919364	Insertion	ins/ins	ins/+	ins/+	1	
1	57314387	57340083	Insertion	+/+	ins/+	ins/+	1	
1	73112725	73112725	Insertion	ins/ins#	ins/ins#	ins/+#	1	
1	73112725	73153143	Insertion	ins/+	ins/+	ins/+	1	
1	74719020	74728535	Insertion	ins/+	ins/+	+/+	1	
1	82251577	82278717	Insertion	ins/ins	ins/ins	ins/ins	1	
1	88504715	88513398	Insertion	ins/ins	ins/ins	ins/ins	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
1	103936432	103943798	Insertion	ins/ins	ins/ins	ins/ins	1	
1	103937109	103943798	Insertion	ins/ins	ins/ins	ins/ins*	1	
1	105947347	105947347	Insertion	ins/+*	+/+	ins/+	1	
1	110117832	110120694	Insertion	ins/ins*	ins/ins	ins/ins	1	
1	110127477	110128963	Insertion	ins/ins	ins/ins	ins/ins	1	
1	120827418	120828465	Insertion	ins/ins	ins/ins	ins/+	1	
1	145407875	145407875	Insertion	ins/ins#	NC	NC	NC	
1	145420350	145420350	Insertion	NC	NC	ins/ins*	NC	
1	146981540	146994357	Insertion	+/+	ins/+	+/+	1	
1	148533217	148580619	Insertion	ins/+#	NC	NC	NC	
1	152206700	152228831	Insertion	ins/+	ins/+	ins/+	1	
1	152467865	152496190	Insertion	+/+	ins/+	+/+	1	
1	161439988	161441931	Insertion	ins/+	ins/+*	NC	NC	
1	165743482	165743482	Insertion	ins/+	+/+	+/+	0	
1	167195010	167212125	Insertion	ins/ins	ins/ins	ins/ins*	1	
1	182290164	182313863	Insertion	ins/+	+/+	ins/ins	1	
1	198477495	198510753	Insertion	ins/+	ins/+*	+/+	1	
1	202198434	202234946	Insertion	ins/+*	ins/+*	ins/+	1	
1	206037899	206068882	Insertion	ins/+	ins/+	ins/+	1	
1	210627183	210656416	Insertion	ins/+*	ins/+	ins/+	1	
1	223080972	223093597	Insertion	ins/+	ins/+	ins/+	1	
1	231786890	231804770	Insertion	ins/+	ins/+	ins/+	1	
1	234201681	234208145	Insertion	ins/+*	ins/+*	ins/ins	1	
1	236095485	236101131	Insertion	ins/ins*	ins/ins*	ins/ins	1	
1	236705614	236707563	Insertion	ins/+*	ins/+	ins/+	1	
1	241961196	241993293	Insertion	ins/+	+/+	ins/+	1	
1	244791050	244804492	Insertion	ins/ins*	ins/+	ins/+	1	
1	245651963	245657588	Insertion	ins/+#	ins/ins#	ins/+#	1	
1	246815292	246821331	Insertion	ins/ins*	ins/+	ins/ins*	1	
1	247355024	247363998	Insertion	ins/+	+/+	ins/+*	1	
1	248403063	248407772	Insertion	ins/ins	ins/ins	ins/ins	1	
2	202934	210503	Insertion	ins/+	ins/+	+/+	1	
2	712090	742625	Insertion	ins/+	ins/+	+/+	1	
2	856565	862638	Insertion	ins/+	ins/+	+/+	1	
2	857206	862638	Insertion	ins/+	ins/+	+/+	1	
2	1218681	1230752	Insertion	ins/ins	NC	NC	1	
2	3044270	3047845	Insertion	ins/ins*	ins/ins	ins/ins*	1	
2	3179097	3181860	Insertion	ins/ins	ins/ins#	ins/ins#	1	
2	5022193	5044268	Insertion	ins/+	ins/+	+/+	1	
2	5487873	5495835	Insertion	ins/ins	ins/ins	ins/ins	1	
2	5488019	5495835	Insertion	ins/ins	ins/ins	ins/ins	1	
2	9403113	9413017	Insertion	+/+	+/+	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
2	9737288	9745990	Insertion	ins/+	+/+	ins/ins	1	
2	9737309	9777056	Insertion	ins/+	+/+	ins/ins	1	
2	10396820	10400450	Insertion	ins/ins	ins/ins	ins/ins	1	
2	22968105	22974184	Insertion	ins/+	ins/+	ins/+	1	DGV
2	22968108	22974196	Insertion	ins/+	ins/+	ins/+	1	DGV
2	24566184	24577144	Insertion	ins/+	+/+	ins/+	1	
2	26336929	26337687	Insertion	ins/ins	ins/ins	ins/ins	1	
2	31820964	31827347	Insertion	ins/ins	ins/ins	ins/ins	1	
2	35654247	35660336	Insertion	ins/+	+/+	ins/+	1	DGV
2	35654251	35660352	Insertion	ins/+	+/+	ins/+	1	DGV
2	36331863	36350967	Insertion	ins/+	ins/+	ins/+	1	
2	47156912	47172022	Insertion	+/+	+/+	ins/+	1	
2	47170743	47172022	Insertion	ins/+	ins/+	ins/ins	1	
2	54927879	54945970	Insertion	ins/ins	ins/ins*	ins/ins	1	
2	57224303	57234061	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
2	57827724	57836524	Insertion	ins/ins	ins/+	ins/ins*	1	
2	57827724	57846775	Insertion	ins/ins	ins/+	ins/ins	1	
2	65518777	65520324	Insertion	ins/ins	ins/+	ins/+	1	
2	67574678	67579680	Insertion	+/+	ins/+	ins/+	1	
2	73779132	73779132	Insertion	ins/+	+/+	ins/+*	1	
2	73783375	73783375	Insertion	ins/+	+/+	ins/+*	1	
2	73798061	73802961	Insertion	ins/+	ins/+	ins/+	1	
2	81870669	81874554	Insertion	ins/+	ins/+	+/+	1	
2	82949084	82962350	Insertion	+/+	+/+	ins/+	1	
2	86928864	86953580	Insertion	ins/+	+/+	ins/+	1	
2	97527075	97527843	Insertion	ins/ins	ins/+	ins/+	1	
2	113947472	113966778	Insertion	ins/+	ins/+	+/+	1	
2	115916600	115941608	Insertion	ins/ins#	ins/+	ins/ins#	1	
2	126308536	126316283	Insertion	+/+	ins/+	+/+	1	
2	129172816	129174820	Insertion	ins/ins	ins/+	ins/ins*	1	
2	130104998	130104998	Insertion	+/+	ins/+*	ins/+	1	
2	143547384	143553448	Insertion	ins/+	ins/+	+/+	1	DGV
2	155668146	155668146	Insertion	ins/+	ins/+	ins/+*	1	
2	155668146	155682227	Insertion	ins/+	ins/+	ins/+	1	
2	157915568	157919494	Insertion	ins/ins	ins/ins	ins/ins	1	
2	166642714	166642714	Insertion	ins/+	ins/+	+/+	1	
2	166642714	166688230	Insertion	ins/+	ins/+	+/+	1	
2	166889178	166903312	Insertion	ins/ins	ins/+	ins/+	1	
2	168861382	168880507	Insertion	ins/+	+/+#	ins/ins#	1	
2	171963943	171986327	Insertion	ins/ins	ins/ins	ins/ins	1	
2	180825764	180838151	Insertion	+/+	ins/+	+/+	1	
2	193667306	193693510	Insertion	ins/+*	ins/+*	ins/ins*	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
2	205335498	205350211	Insertion	ins/+	ins/ins*	ins/+*	1	
2	206217255	206229908	Insertion	+/+	ins/+	ins/+	1	
2	208077757	208128027	Insertion	ins/+*	ins/+	ins/ins	1	
2	213270834	213336054	Insertion	ins/+*	ins/+*	ins/+*	1	
2	231820031	231874626	Insertion	+/+	ins/+	ins/+*	1	
2	237631290	237656888	Insertion	ins/+*	ins/+	ins/+	1	
2	239637240	239646781	Insertion	ins/+	+/+	ins/+	1	
2	240922738	240933269	Insertion	ins/+	+/+	ins/+	1	
2	241573469	241597645	Insertion	ins/ins	ins/ins	ins/ins*	1	
2	241804320	241814517	Insertion	+/+	+/+	ins/+	1	
2	241885992	241897354	Insertion	ins/ins	ins/ins	ins/ins	1	
2	241886538	241897354	Insertion	ins/ins	ins/ins	ins/ins	1	
2	241897354	241897354	Insertion	ins/ins	ins/ins	ins/ins	1	
3	12634716	12663211	Insertion	ins/+	ins/+	ins/+	1	
3	18784540	18793066	Insertion	ins/+*	ins/+*	ins/+*	1	
3	20287425	20305090	Insertion	ins/+	ins/+	ins/+	1	
3	20685600	20713288	Insertion	ins/+	+/+	ins/+	1	
3	20713288	20713288	Insertion	ins/+*	+/+	ins/+	1	
3	32932769	32946550	Insertion	ins/ins	ins/ins	ins/ins*	1	
3	37706852	37709589	Insertion	ins/ins	ins/ins	ins/ins*	1	
3	38080665	38088054	Insertion	ins/+*	ins/+*	ins/+	1	
3	38568459	38588285	Insertion	ins/+	ins/+	ins/+	1	
3	47183590	47226781	Insertion	+/+	ins/+	+/+	1	
3	47782114	47804224	Insertion	ins/ins	ins/ins	ins/ins#	1	
3	55754481	55759841	Insertion	ins/ins	ins/ins#	ins/+#	1	
3	71428119	71436169	Insertion	ins/ins	ins/ins	ins/ins	1	
3	75395926	75415136	Insertion	ins/ins	ins/ins*	ins/ins*	1	
3	75395926	75417869	Insertion	ins/ins	ins/ins*	ins/ins*	1	
3	80528488	80555746	Insertion	+/+	+/+	ins/+	1	
3	84384213	84390305	Insertion	ins/+*	+/+	ins/+	1	DGV
3	84895416	84905971	Insertion	ins/ins	ins/+	ins/+	1	
3	84905971	84905971	Insertion	ins/ins	ins/+	ins/+	1	
3	85519673	85531158	Insertion	ins/+	+/+	ins/+	1	
3	85519673	85532619	Insertion	ins/+	+/+	ins/+	1	
3	86215675	86223267	Insertion	+/+	ins/+	+/+	1	
3	88961918	88989505	Insertion	ins/ins*	ins/ins	ins/+	1	
3	94891377	94944610	Insertion	ins/+#	ins/ins#	ins/+	1	
3	101549599	101565040	Insertion	ins/+	ins/+	+/+	1	
3	112163989	112164759	Insertion	+/+	ins/+	ins/+	1	
3	112164374	112166955	Insertion	+/+	ins/+	ins/+	1	
3	123871289	123875514	Insertion	ins/ins	ins/+	ins/ins	1	
3	128123351	128141189	Insertion	ins/+	ins/+	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
3	143398240	143410558	Insertion	ins/+	+/+	ins/ins*	1	
3	145268203	145274535	Insertion	ins/ins	ins/+	ins/ins	1	
3	151422041	151430054	Insertion	ins/+	ins/ins	ins/+	1	
3	151430054	151430054	Insertion	ins/+	ins/ins#	ins/+	1	
3	151430738	151436811	Insertion	ins/+	ins/ins*	ins/+	1	DGV
3	151664469	151675585	Insertion	ins/+	+/+	ins/+	1	
3	152689271	152696147	Insertion	ins/ins	ins/ins	ins/ins	1	
3	154172724	154184477	Insertion	ins/+	ins/ins*	ins/+	1	
3	163368687	163368687	Insertion	+/+	ins/+*	ins/+*	1	
3	175359869	175392576	Insertion	ins/+#	ins/+#	ins/+#	1	
3	176218298	176259574	Insertion	+/+	+/+	ins/+	1	
3	181471021	181475088	Insertion	ins/ins*	ins/+	ins/+	1	
3	184431292	184443864	Insertion	ins/ins*	ins/+#	ins/ins	1	
3	186654310	186660389	Insertion	ins/+	+/+	ins/+	1	DGV
3	193200898	193201888	Insertion	+/+	ins/+	ins/+	1	
3	195442428	195453967	Insertion	ins/ins	ins/ins	ins/ins	1	
3	195471626	195513125	Insertion	ins/+#	ins/+#	ins/+#	1	
3	195642970	195644278	Insertion	+/+	+/+	ins/+	1	
3	195643482	195643482	Insertion	+/+	+/+	ins/+	1	
3	195704768	195716598	Insertion	ins/+	ins/+	+/+	1	
3	195715961	195746345	Insertion	ins/+	ins/+	+/+	1	
3	195784848	195789845	Insertion	ins/+*	ins/+	ins/+	1	
3	195785240	195789845	Insertion	+/+*	ins/+*	ins/+*	1	
3	197381564	197394910	Insertion	ins/ins	ins/ins	ins/ins*	1	
3	197381564	197470209	Insertion	ins/+	ins/+	ins/ins*	1	
4	545064	569896	Insertion	ins/+*	ins/+*	ins/ins	1	
4	1394335	1416658	Insertion	ins/+	ins/+	+/+	1	
4	8626396	8632033	Insertion	+/+	ins/+	ins/+*	1	
4	8627088	8632726	Insertion	ins/+	ins/+	ins/ins	1	
4	8632033	8636998	Insertion	ins/+	+/+	ins/+*	1	
4	9702170	9705217	Insertion	ins/+	+/+	ins/+	1	
4	14904063	14926887	Insertion	ins/+*	ins/+#	ins/ins	1	
4	16318555	16333562	Insertion	ins/+	ins/+	+/+	1	
4	21593405	21611345	Insertion	ins/+	+/+	ins/+	1	
4	37576847	37613322	Insertion	ins/+	ins/+	+/+	1	
4	40282086	40300382	Insertion	ins/ins*	ins/ins*	ins/+	1	
4	55725585	55748842	Insertion	ins/+	ins/+	ins/+	1	
4	56870931	56877152	Insertion	ins/+	ins/ins	+/+	1	
4	68707293	68725415	Insertion	ins/+	ins/+	ins/+	1	
4	70089872	70114851	Insertion	ins/+	ins/+	ins/+	1	
4	73264122	73272426	Insertion	ins/+	ins/+	+/+	1	
4	87915173	87926488	Insertion	ins/+*	+/+*	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
4	88163549	88205279	Insertion	ins/ins*	ins/+*	ins/+	1	
4	91037181	91049565	Insertion	ins/ins	ins/ins	ins/ins	1	
4	96500882	96506562	Insertion	+/+	+/+	ins/+	1	
4	111702972	111711100	Insertion	+/+	+/+	ins/+	1	
4	131260461	131266564	Insertion	ins/+	ins/+	ins/+	1	DGV
4	131260475	131266568	Insertion	ins/+	ins/+	ins/+	1	DGV
4	131708035	131788516	Insertion	ins/+#	ins/+#	ins/ins#	1	
4	137885038	137902639	Insertion	ins/+	ins/ins	ins/+	1	
4	146280916	146314533	Insertion	ins/+	+/+	ins/+	1	
4	150247864	150255410	Insertion	ins/+	+/+	ins/ins	1	
4	150247864	150258545	Insertion	ins/+	ins/+	+/+	1	
4	151809866	151810702	Insertion	+/+	ins/+	+/+	1	
4	154148300	154166947	Insertion	ins/ins	ins/ins	ins/ins*	1	
4	154148685	154166947	Insertion	ins/ins	ins/ins	NC	NC	
4	155953347	155964750	Insertion	ins/ins	ins/ins	ins/ins	1	
4	166571510	166583927	Insertion	ins/+	ins/+	ins/ins	1	
4	181238751	181245113	Insertion	ins/+	+/+	ins/+	1	
4	181238751	181306022	Insertion	ins/+	ins/ins	ins/+	1	
4	182827329	182850268	Insertion	ins/+*	ins/+	ins/+	1	
4	186418310	186438617	Insertion	ins/+	+/+	ins/ins	1	
4	186419907	186438617	Insertion	ins/+	+/+	ins/+	1	
4	188502726	188518627	Insertion	ins/ins	ins/ins	ins/+	1	
4	189224891	189245331	Insertion	ins/+	ins/ins	+/+	1	
4	189305700	189314111	Insertion	ins/+	ins/+*	+/+	1	
4	189551222	189560086	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
4	189822550	189882021	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
4	189882021	189882021	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
5	61588	115860	Insertion	ins/+	ins/+	ins/+	1	
5	647125	662300	Insertion	+/+	ins/+	ins/+	1	
5	1331824	1342492	Insertion	ins/+	ins/+	ins/ins	1	
5	1331824	1416587	Insertion	ins/ins*	ins/+	ins/ins	1	
5	1409770	1416587	Insertion	ins/+	ins/ins*	ins/+*	1	
5	2143914	2147882	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
5	2143914	2151448	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
5	3319942	3325010	Insertion	ins/+*	ins/+*	ins/ins#	1	
5	5304142	5319232	Insertion	+/+	ins/+	ins/+	1	
5	7259613	7271758	Insertion	ins/ins	ins/+*	ins/+	1	
5	7370070	7376568	Insertion	+/+	ins/+	+/+	1	
5	17709913	17712826	Insertion	ins/+	+/+	ins/+	1	
5	21197711	21214962	Insertion	ins/ins	ins/+	ins/+	1	
5	21471613	21480997	Insertion	ins1/ins1	ins1/ins2	ins1/ins3	1	
5	21480997	21496651	Insertion	NC	ins/ins	NC	NC	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
5	29065894	29071801	Insertion	ins/ins	ins/+	ins/ins	1	
5	33624275	33643275	Insertion	ins/+	ins/+	ins/ins	1	
5	33796765	33796765	Insertion	ins/+	ins/+	ins/+	1	
5	33797414	33803522	Insertion	ins/+	ins/+	ins/+	1	DGV
5	33797424	33803488	Insertion	ins/+	ins/+	ins/+	1	DGV
5	56376724	56381386	Insertion	ins/+	ins/+	+/+	1	
5	71202792	71216808	Insertion	NC	ins/+#	+/+*	NC	
5	76345841	76363289	Insertion	+/+	ins/+	+/+	1	
5	83059449	83066798	Insertion	ins/+	ins/ins	ins/+	1	
5	90154934	90161053	Insertion	ins/ins	ins/ins	ins/ins	1	DGV
5	95210391	95227942	Insertion	ins/+	ins/+	ins/+	1	
5	95227942	95227942	Insertion	ins/ins	ins/ins	ins/ins	1	
5	95227942	95233841	Insertion	ins/+	ins/+	ins/+	1	
5	99862830	99874669	Insertion	ins/+	+/+	ins/+	1	
5	99863480	99874669	Insertion	ins/+	+/+	ins/+	1	
5	110142195	110179322	Insertion	ins/ins	ins/ins	ins/+*	1	
5	119088598	119140682	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
5	140370687	140382578	Insertion	+/+	+/+	ins/+	1	
5	141184656	141184656	Insertion	+/+	ins/+	ins/+	1	
5	141184656	141187934	Insertion	+/+	ins/+	ins/+	1	
5	141983173	142009928	Insertion	ins/+*	ins/+	ins/ins*	1	
5	153935776	153954501	Insertion	+/+	+/+	ins/+	1	
5	162527571	162532165	Insertion	ins/+	ins/+	ins/+#	1	
5	177908894	177928173	Insertion	ins/ins	ins/ins	ins/ins	1	
5	177908894	177928564	Insertion	ins/+#	ins/ins#	ins/+*	1	
5	178583020	178595280	Insertion	ins2/ins2	ins2/ins2	ins1/ins2	1	
5	181032276	181049054	Insertion	ins/ins	ins/ins	ins/ins	1	
5	181317467	181326681	Insertion	ins/ins	ins/ins	ins/+	1	
5	181317467	181327212	Insertion	ins/ins*	ins/ins*	ins/+	1	
5	181433557	181440729	Insertion	ins/+	ins/+	ins/+	1	
6	364330	383892	Insertion	ins/+	ins/+	ins/+*	1	
6	13122406	13134963	Insertion	ins/ins	ins/ins	ins/ins	1	
6	13134963	13134963	Insertion	ins/ins	ins/ins	ins/ins	1	
6	13190761	13196838	Insertion	+/+	+/+	ins/+	1	DGV
6	13190767	13196839	Insertion	+/+	+/+	ins/+	1	DGV
6	13494182	13494182	Insertion	ins/ins	ins/ins	ins/ins	1	
6	13494182	13499469	Insertion	ins/ins	ins/ins	ins/ins	1	
6	25063682	25087495	Insertion	ins/+	ins/+	ins/+	1	
6	32383843	32388120	Insertion	NC	+/+	ins/ins	NC	
6	35782171	35782171	Insertion	+/+	+/+	ins/+	1	
6	35787822	35795009	Insertion	ins/+	ins/+	ins/+	1	
6	47459837	47478414	Insertion	ins/+	ins/ins*	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
6	64291061	64299891	Insertion	ins/+	ins/ins	ins/+	1	
6	65840036	65859253	Insertion	ins/+	ins/ins	ins/+	1	
6	65840036	65859657	Insertion	ins/+	ins/ins	ins/+	1	
6	67608595	67613004	Insertion	ins/+	ins/+	ins/+	1	
6	67613004	67613004	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
6	69562912	69569905	Insertion	ins/ins*	ins/ins*	ins/+*	1	
6	72087831	72100103	Insertion	+/+	ins/+	+/+	1	
6	79357672	79366997	Insertion	ins/+	ins/+	ins/ins	1	
6	79366613	79377104	Insertion	ins/+	ins/+	ins/ins	1	
6	79896289	79910882	Insertion	del/+	del/+	ins/+	1	
6	82255936	82281778	Insertion	ins/ins#	ins/+#	ins/ins#	1	
6	85924254	85934771	Insertion	ins/+	ins/+	+/+	1	
6	90954097	90967280	Insertion	ins/+	ins/ins	ins/+	1	
6	94029101	94079186	Insertion	ins/+*	ins/+*	ins/+	1	
6	102390401	102399112	Insertion	ins/+	ins/+	+/+	1	
6	102390977	102399112	Insertion	ins/+	ins/+	+/+	1	
6	110581502	110611253	Insertion	+/+	+/+	ins/+	1	
6	123532773	123538838	Insertion	ins/+	ins/+	+/+	1	DGV
6	127999358	128011380	Insertion	NC	ins/+	ins/+	NC	
6	142128909	142130107	Insertion	ins/+	+/+	ins/ins	1	
6	147842416	147848518	Insertion	+/+	+/+	ins/+	1	DGV
6	148735405	148740378	Insertion	ins/ins	ins/ins	ins/ins	1	
6	157304022	157321680	Insertion	ins/+#	ins/ins#	ins/+#	1	
6	157533988	157553647	Insertion	ins/ins*	ins/+	ins/ins*	1	
6	157553647	157553647	Insertion	ins/ins	ins/+	ins/ins	1	
6	160212439	160220735	Insertion	ins/ins	ins/ins	ins/ins	1	
6	160774937	160787823	Insertion	+/+	ins/+	ins/+	1	
6	161753940	161763503	Insertion	ins/+	ins/ins	+/+	1	
6	168225504	168234338	Insertion	ins/ins*	ins/ins*	ins/+	1	
6	168583741	168613459	Insertion	ins/+*	ins/+	+/+	1	
6	170139862	170148997	Insertion	ins/ins*	ins/+*	ins/ins	1	
6	170373208	170378903	Insertion	+/+	ins/+	ins/+	1	
6	170397873	170402448	Insertion	ins/ins*	ins/+	ins/+	1	
6	170709153	170716467	Insertion	+/+	ins/+	ins/+	1	
7	903196	927587	Insertion	ins/ins*	ins/ins	ins/ins*	1	
7	1193613	1197327	Insertion	ins1/ins2	ins1/ins2	ins1/ins1	1	
7	1271955	1277783	Insertion	ins/ins	ins/ins	ins/+	1	
7	7973569	7975100	Insertion	ins/+	+/+	ins/+	1	
7	7979358	7985404	Insertion	ins/+	+/+	ins/+	1	DGV
7	7979362	7985444	Insertion	ins/+	+/+	ins/+	1	DGV
7	9022635	9082381	Insertion	ins/+	ins/+	ins/+	1	
7	19470641	19488514	Insertion	ins/ins*	ins/ins*	ins/ins*	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
7	23482217	23530976	Insertion	ins/+#	ins/+#	ins/+#	1	
7	42490249	42496332	Insertion	ins/+	ins/+	ins/+	1	DGV
7	54305873	54309747	Insertion	ins/+*	ins/+*	ins/+*	1	
7	56803644	57051656	Insertion	ins/+	NC	NC	NC	
7	57038066	57051656	Insertion	ins/+	+/+	ins/ins	1	
7	63030201	63035649	Insertion	ins/ins	ins/+	ins/+	1	
7	63745279	63772776	Insertion	+/+	ins/+#	+/+	1	
7	63748024	63753047	Insertion	+/+	+/+	ins/+	1	
7	65481931	65512830	Insertion	ins1/del	ins1/ins2	ins1/del	1	
7	65514658	65530747	Insertion	ins/+#	ins/+#	ins/+#	1	
7	66790009	66799577	Insertion	+/+	ins/+	+/+	1	
7	67115708	67125176	Insertion	ins/ins	ins/ins	ins/+	1	
7	71676694	71688269	Insertion	ins/+	ins/+	+/+	1	
7	72791213	72806996	Insertion	ins/+	ins/ins	ins/+	1	
7	75365332	75365332	Insertion	ins/+#	ins/+#	+/+	1	
7	76494140	76500339	Insertion	ins/ins	ins/ins	ins/ins	1	
7	99200358	99233703	Insertion	ins/ins	ins/ins#	ins/ins*	1	
7	100949356	101042140	Insertion	ins/ins	ins/ins	ins/ins	1	
7	100949356	100994113	Insertion	+/+	+/+	ins/+	1	
7	100972909	100994113	Insertion	ins/ins	ins/ins	ins/+	1	
7	103158152	103172695	Insertion	+/+	ins/+	+/+	1	
7	107732580	107734197	Insertion	ins/ins	ins/ins	ins/ins	1	
7	108171984	108174750	Insertion	ins/+	ins/+	ins/ins	1	
7	108188786	108194896	Insertion	ins/+	ins/+	ins/+	1	DGV
7	108188787	108194868	Insertion	ins/+	ins/+	ins/+	1	DGV
7	135287858	135292945	Insertion	ins/+	ins/+	ins/+	1	
7	142351595	142351595	Insertion	ins/+	ins/+	+/+	1	
7	142355487	142359034	Insertion	+/+	ins/+	+/+	1	
7	142519698	142525528	Insertion	ins/ins	ins/ins	ins/ins	1	
7	142519698	142519698	Insertion	ins/ins	ins/ins	ins/ins	1	
7	142541644	142768532	Insertion	ins/ins*	ins/+*	ins/ins*	1	
7	142757956	142768532	Insertion	ins/ins	ins/ins	ins/+	1	
7	142768532	142768532	Insertion	ins/ins*	ins/ins*	ins/+	1	
7	151053699	151062927	Insertion	ins/+	ins/+	ins/+	1	
7	151053699	151067013	Insertion	ins/+	ins/+	ins/+	1	
7	155313831	155340069	Insertion	ins/ins	ins/ins*	ins/ins*	1	
7	155313831	155353458	Insertion	ins/ins	ins/ins*	ins/ins*	1	
7	155890777	155893413	Insertion	ins/ins*	ins/ins	ins/ins	1	
7	158143103	158160589	Insertion	ins/+	ins/+	ins/+	1	
7	158583654	158596917	Insertion	ins/+	ins/+	ins/ins*	1	
7	158583682	158596917	Insertion	ins/ins	ins/ins	ins/ins	1	
7	158603153	158604500	Insertion	ins/+	ins/+	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
7	159190946	159205819	Insertion	ins/+	ins/+	ins/+	1	
7	159194140	159216182	Insertion	ins/+	ins/+	ins/+	1	
8	986131	989573	Insertion	ins/+	ins/+	ins/ins	1	
8	2183568	2194509	Insertion	ins1/+	ins1/ins2	ins2/+	1	
8	2184094	2194509	Insertion	ins/+	ins/ins	ins/+	1	
8	2381675	2390307	Insertion	+/+	ins/+	+/+	1	
8	2464906	2480998	Insertion	ins/+	+/+	ins/+*	1	
8	21043017	21050939	Insertion	ins/ins	ins/ins	ins/ins	1	
8	25068811	25073234	Insertion	+/+	+/+	ins/+	1	
8	34440191	34447939	Insertion	ins/ins	ins/ins	ins/ins	1	
8	43234107	43253844	Insertion	ins/ins*	ins/ins*	ins/+	1	
8	46631424	46632194	Insertion	ins/+	+/+	ins/+	1	
8	49711307	49714946	Insertion	ins/ins*	ins/ins	ins/ins*	1	
8	57067558	57077027	Insertion	ins/ins	ins/ins	ins/ins	1	
8	61204185	61217404	Insertion	ins1/ins2	ins1/ins2	ins2/+	1	
8	85775260	85790535	Insertion	ins/+*	ins/+*	ins/ins*	1	
8	102538489	102553854	Insertion	ins/+	ins/ins	ins/+	1	
8	120913953	120921083	Insertion	ins/+	ins/+	+/+	1	
8	129734346	129747367	Insertion	+/+	+/+	ins/+	1	
8	129735096	129747367	Insertion	+/+	+/+	ins/+	1	
8	130424466	130430775	Insertion	+/+	+/+	ins/+	1	
8	141414607	141424738	Insertion	ins/ins	ins/ins	ins/ins	1	
8	141630340	141638024	Insertion	ins/ins	ins/ins	ins/+	1	
8	141751951	141779870	Insertion	ins/ins	ins/ins	ins/ins	1	
8	141772332	141779870	Insertion	ins/ins	ins/ins	ins/ins	1	
8	143214389	143250012	Insertion	ins/+*	ins/+*	ins/ins*	1	
8	143856843	143872031	Insertion	ins/+	ins/ins	+/+	1	
8	144115589	144128984	Insertion	ins/ins	ins/ins	ins/ins	1	
8	144676102	144691084	Insertion	ins/+	ins/+*	ins/+*	1	
9	4254519	4282544	Insertion	+/+	ins/+	+/+	1	
9	5478356	5493436	Insertion	+/+	+/+	ins/+	1	
9	5493436	5493436	Insertion	+/+	+/+	ins/+	1	
9	6377928	6405678	Insertion	ins/+	ins/ins	ins/+	1	
9	9003774	9018403	Insertion	ins/ins*	ins/ins#	ins/ins#	1	
9	10793419	10802646	Insertion	+/+	+/+	ins/+	1	
9	35912870	35922565	Insertion	ins/+	ins/+#	ins/+	1	
9	39811724	39814487	Insertion	ins/+	ins/+	ins/+	1	
9	40812477	40816882	Insertion	ins/+	ins/+	ins/+	1	
9	40812477	40876207	Insertion	ins/+	ins/+	ins/+	1	
9	41632934	41655531	Insertion	ins/+	ins/ins	ins/+	1	
9	41644888	41647910	Insertion	ins/+	ins/ins	ins/+	1	
9	42140851	42349392	Insertion	ins/+	ins/+	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
9	63740990	63775277	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
9	66747178	66755255	Insertion	ins/+	ins/+	+/+	1	
9	67328781	67359854	Insertion	ins/+*	ins/ins*	ins/+*	1	
9	67645154	67672963	Insertion	ins/+	ins/ins*	ins/+#	1	
9	67672963	67672963	Insertion	ins/ins	ins/ins	ins/ins	1	
9	70235668	70242733	Insertion	ins/+	ins/+	+/+	1	
9	70236053	70236053	Insertion	ins/+	ins/+	+/+	1	
9	77153743	77166700	Insertion	ins/ins	ins/+	ins/+	1	
9	79464477	79474638	Insertion	ins/+	ins/+	+/+	1	
9	88193934	88211143	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
9	88223574	88224877	Insertion	ins/ins	ins/ins	ins/ins	1	
9	88656524	88664534	Insertion	ins/ins	ins/ins	ins/ins	1	
9	91641731	91655475	Insertion	ins/+	+/+	ins/+	1	
9	105074990	105095584	Insertion	+/+	+/+	ins/+	1	
9	113059064	113059064	Insertion	ins/+*	ins/+*	ins/+*	1	
9	113059064	113063848	Insertion	ins/+	ins/+	+/+	1	
9	113085632	113094701	Insertion	ins1/ins1	ins1/ins1	ins1/ins2	1	
9	114315287	114319416	Insertion	+/+	+/+	ins/+	1	
9	114315287	114320130	Insertion	+/+	+/+	ins/+	1	
9	118629287	118652059	Insertion	ins/+	ins/+	ins/+	1	
9	125805091	125816295	Insertion	ins/ins	ins/ins	ins/ins	1	
9	125805188	125816295	Insertion	ins/ins	ins/ins	ins/ins	1	
9	130956712	130960940	Insertion	ins/ins	ins/ins	ins/ins	1	
9	131406288	131416081	Insertion	ins/+	ins/+	ins/ins	1	
9	131736176	131742900	Insertion	+/+	+/+	ins/+*	1	
9	131750329	131756096	Insertion	+/+	+/+	ins/+	1	
9	132943867	132944752	Insertion	ins/+	ins/ins	+/+	1	
9	132949319	132956524	Insertion	ins/+	ins/ins	+/+	1	
9	135333853	135350287	Insertion	ins/+	ins/+	+/+	1	
9	137223236	137354372	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
9	137324583	137333089	Insertion	ins/+	ins/+	ins/+	1	
9	137324583	137334650	Insertion	ins/+	ins/+	ins/+	1	
9	137324936	137333089	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
9	137499412	137521642	Insertion	ins/+*	ins/+#	ins/+#	1	
9	137711264	137724336	Insertion	ins/+	ins/+	ins/+	1	
9	137711644	137724336	Insertion	ins/ins	ins/ins	ins/ins	1	
9	137712196	137724336	Insertion	ins/+	ins/+	ins/+	1	
9	138274566	138279726	Insertion	ins/+#	ins/ins#	ins/+#	1	
10	1225133	1244692	Insertion	ins/+	+/+	ins/+	1	
10	1225514	1244692	Insertion	ins/+	+/+	ins/+	1	
10	2382641	2393676	Insertion	ins/+	+/+	ins/+	1	
10	8805802	8808035	Insertion	+/+	+/+	ins/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
10	12772891	12776318	Insertion	ins/ins	ins/+	ins/+	1	
10	15190387	15210783	Insertion	ins/ins	ins/ins	ins/ins*	1	
10	25385442	25450293	Insertion	ins/ins	ins/+	ins/ins	1	
10	26892789	26899162	Insertion	ins/ins	ins/ins	ins/ins	1	
10	27300900	27319775	Insertion	ins/+	ins/+	+/+	1	
10	27301285	27319775	Insertion	ins/+	ins/+*	+/+	1	
10	36447175	36458767	Insertion	ins/+	+/+	ins/+*	1	
10	36464550	36482055	Insertion	ins/+	NC	NC	NC	
10	36464550	36464550	Insertion	ins/+	+/+	ins/+*	1	
10	42268659	42280510	Insertion	ins/+	ins/+	ins/ins#	1	
10	45338337	45343900	Insertion	ins/+	+/+	ins/+*	1	
10	46273708	46286951	Insertion	ins/+	ins/+	ins/ins	1	
10	48052162	48064411	Insertion	ins/+	+/+	+/+	0	
10	56768390	56807967	Insertion	ins/+	ins/+	+/+	1	
10	65519415	65530624	Insertion	ins/ins	ins/ins*	ins/+	1	
10	83127888	83133944	Insertion	ins/+	ins/ins	+/+	1	
10	83206191	83219312	Insertion	+/+	ins/+	+/+	1	
10	86495006	86505818	Insertion	ins/ins	ins/ins	ins/ins	1	
10	89032120	89051675	Insertion	ins/+	ins/+	+/+	1	
10	109198786	109215909	Insertion	+/+	ins/+	+/+	1	
10	122695681	122701747	Insertion	ins/+	ins/+	ins/+	1	DGV
10	122695684	122701769	Insertion	ins/+	ins/+	ins/+	1	DGV
10	123252517	123263144	Insertion	ins/ins	ins/+*	ins/ins*	1	
10	126939190	126956197	Insertion	ins/ins	ins/ins	ins/ins	1	
10	128463139	128464756	Insertion	ins1/ins2	ins1/ins1	ins2/ins2	1	
10	131039748	131061103	Insertion	ins/+#	ins/+#	+/+#	1	
10	132151511	132175328	Insertion	ins/ins	ins/ins	ins/ins	1	
10	132355848	132375328	Insertion	ins/+	ins/+	ins/ins	1	
11	174006	184906	Insertion	ins/+	+/+	ins/+	1	
11	174006	199516	Insertion	ins/+	ins/+	ins/+	1	
11	336372	366388	Insertion	ins/+*	ins/+*	ins/ins*	1	
11	399027	407456	Insertion	ins/+	ins/ins	ins/+	1	
11	576376	585983	Insertion	ins/ins	NC	NC	NC	
11	581475	585983	Insertion	ins/ins	NC	NC	NC	
11	993046	1003070	Insertion	ins/ins	ins/ins	ins/+*	1	
11	995232	1003070	Insertion	ins/ins	ins/ins	ins/+*	1	
11	1011887	1019624	Insertion	ins/+	ins/+	ins/+	1	
11	1077810	1102584	Insertion	ins/ins*	ins/+*	ins/ins*	1	
11	1092262	1102584	Insertion	ins/ins	ins/ins	ins/ins	1	
11	1536352	1536352	Insertion	ins/ins	ins/ins	ins/ins	1	
11	1536352	1541923	Insertion	ins/+*	ins/+#	ins/+#	1	
11	1596369	1608938	Insertion	ins/+*	ins/ins	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
11	1927077	1943801	Insertion	ins/+*	ins/+	ins/+#	1	
11	1930475	1943801	Insertion	ins/+*	ins/+*	ins/+#	1	
11	3642907	3644532	Insertion	ins/+	ins/+	+/+	1	
11	3644532	3658522	Insertion	ins/+	ins/+	+/+	1	
11	7765808	7838292	Insertion	ins/ins	ins/ins	ins/ins*	1	
11	7813635	7840033	Insertion	ins/ins#	ins/ins	ins/+#	1	
11	11235386	11263316	Insertion	ins/ins	ins/ins	ins/ins	1	
11	12233699	12243117	Insertion	+/+	+/+	ins/+*	1	
11	12609816	12641630	Insertion	ins/ins	ins/ins	ins/+	1	
11	12611092	12641630	Insertion	ins/ins	ins/ins	ins/+	1	
11	14260402	14292505	Insertion	ins/+	ins/+	ins/+	1	
11	23322629	23367042	Insertion	ins/+*	ins/ins*	ins/+	1	
11	24374923	24381570	Insertion	+/+	ins/+	ins/+#	1	
11	25177274	25250231	Insertion	ins/+#	ins/+#	ins/+#	1	
11	42944631	42954558	Insertion	+/+	ins/+	+/+	1	
11	47637438	47646702	Insertion	ins/ins	ins/ins	ins/ins	1	
11	47889153	47894048	Insertion	ins/ins	ins/ins	ins/ins	1	
11	48903103	48903103	Insertion	ins/ins*	ins/ins*	ins/+*	1	
11	48909340	48948355	Insertion	ins/ins*	ins/ins	ins/+#	1	
11	50144245	50144245	Insertion	ins/+	+/+	ins/+*	1	
11	56373561	56390187	Insertion	ins/ins	ins/ins	ins/ins	1	
11	56694059	56698202	Insertion	ins/ins	ins/ins	ins/ins	1	
11	56698202	56713860	Insertion	ins/ins	ins/ins	ins/ins	1	
11	60031511	60061239	Insertion	ins/+*	ins/+	ins/+	1	
11	64535380	64560160	Insertion	ins/ins	ins/ins	ins/ins	1	
11	67829615	67835785	Insertion	+/+	+/+	ins/+	1	
11	68869976	68898521	Insertion	ins/ins	ins/ins	ins/ins	1	
11	69165612	69187527	Insertion	ins/ins	ins/ins	ins/ins	1	
11	69173822	69187527	Insertion	ins/ins	ins/ins	ins/ins	1	
11	70364362	70373331	Insertion	ins/+	ins/+	ins/+	1	
11	70364898	70373331	Insertion	ins/+#	NC	NC	NC	
11	90024201	90040508	Insertion	ins/+	+/+	ins/+	1	
11	90257841	90269007	Insertion	ins/+#	+/+	ins/+#	1	
11	92306681	92332036	Insertion	ins/+*	ins/+*	ins/+*	1	
11	93903641	94236619	Insertion	ins/+	ins/+	ins/+	1	
11	94236234	94237004	Insertion	ins/ins	ins/ins	ins/ins	1	
11	94237004	94244039	Insertion	ins/+*	ins/+	ins/+*	1	
11	110507132	110513223	Insertion	ins/+	ins/+	ins/+	1	DGV
11	115675371	115678174	Insertion	ins/+	ins/+	ins/+	1	
12	30981	40142	Insertion	ins/+	+/+	ins/+	1	
12	1072349	1075108	Insertion	+/+	ins/+	+/+	1	
12	1305967	1325127	Insertion	+/+	ins/+	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
12	2254969	2257962	Insertion	ins/ins	ins/+	ins/+	1	
12	5908535	5934840	Insertion	ins/ins*	ins/ins	ins/ins	1	
12	6140344	6148195	Insertion	ins/+	ins/ins	ins/+	1	
12	6955355	6960920	Insertion	ins/ins	ins/+	ins/ins	1	
12	8227147	8227147	Insertion	+/+	ins/+	+/+	1	
12	8227147	8230313	Insertion	+/+	ins/+	+/+	1	
12	11403761	11408442	Insertion	+/+	+/+	ins/+*	1	
12	12644069	12669828	Insertion	ins/ins#	NC	NC	NC	
12	12780727	12786789	Insertion	ins/+*	NC	NC	1	DGV
12	25620151	25654555	Insertion	+/+	+/+	ins/+	1	
12	28062106	28082859	Insertion	ins/+	+/+	ins/ins	1	
12	33850048	33865597	Insertion	ins/ins	ins/ins	ins/+	1	
12	33850433	33865597	Insertion	ins/ins	ins/ins	ins/+	1	
12	40467256	40494443	Insertion	ins/+	ins/+	+/+	1	
12	40494443	40494443	Insertion	ins/+	ins/ins	+/+	1	
12	40500913	40513773	Insertion	+/+*	ins/+	+/+	1	
12	55344277	55344277	Insertion	ins/+	ins/+	ins/ins	1	
12	55344277	55354526	Insertion	ins/+	ins/+	ins/ins	1	
12	58074483	58076955	Insertion	ins/ins	ins/ins	ins/ins	1	
12	58632116	58635796	Insertion	ins/+	ins/ins	ins/+	1	
12	70933732	70963372	Insertion	ins/+	+/+	ins/ins	1	
12	76118227	76135035	Insertion	ins/ins	ins/ins	ins/ins#	1	
12	78596850	78605413	Insertion	ins/+	ins/+	ins/+	1	
12	78605413	78605413	Insertion	ins/+	ins/+	ins/+	1	
12	79760257	79769683	Insertion	ins/+	ins/+	ins/ins	1	
12	86246134	86263158	Insertion	ins/ins	ins/ins	ins/+	1	
12	92910660	92917265	Insertion	ins/+	+/+	ins/ins	1	
12	101147571	101157445	Insertion	ins/+	+/+	ins/+	1	
12	101147956	101157445	Insertion	ins/+	+/+	ins/+	1	
12	107912984	107916022	Insertion	ins/ins	ins/+	ins/+	1	
12	108567826	108582891	Insertion	ins/ins	ins/ins	ins/ins	1	
12	108577946	108582891	Insertion	ins/ins	ins/ins	ins/ins	1	
12	117376624	117382708	Insertion	ins/+	ins/+	ins/+	1	DGV
12	118862643	118867504	Insertion	+/+	ins/+	ins/+	1	
12	125309660	125312911	Insertion	ins/ins	ins/+	ins/+	1	
12	126322874	126332042	Insertion	ins/+	ins/+	+/+	1	
12	126823263	126843639	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
12	127149743	127166801	Insertion	+/+	ins/+	ins/+	1	
12	130645446	130650386	Insertion	ins/+	ins/+	ins/+	1	
12	132147379	132155124	Insertion	+/+	ins/+	+/+	1	
12	132949971	132967479	Insertion	ins/+#	ins/ins	NC	1	
13	21369771	21381557	Insertion	ins/+	ins/+	ins/ins	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
13	21724343	21730375	Insertion	ins/ins*	ins/ins	ins/ins*	1	
13	25799617	25821741	Insertion	ins/ins	ins/ins	ins/+	1	
13	27060305	27066359	Insertion	ins/+	ins/+	ins/+*	1	
13	35950827	35967132	Insertion	ins/+*	ins/ins	ins/+*	1	
13	38955355	38958112	Insertion	ins/ins	ins/ins	ins/ins	1	
13	46582843	46590316	Insertion	ins/+	+/+	ins/+	1	
13	52530686	52539973	Insertion	ins/+	ins/+	+/+	1	
13	57074942	57179234	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
13	60888180	60894256	Insertion	ins/ins	ins/ins	ins/+	1	DGV
13	60888180	60894291	Insertion	ins/ins	ins/ins	ins/+	1	DGV
13	63375243	63388602	Insertion	ins/ins*	ins/+	ins/+	1	
13	84325422	84408682	Insertion	+/+	ins/+	+/+	1	
13	89648423	89686861	Insertion	ins/+#	ins/+#	ins/+#	1	
13	106772227	106782349	Insertion	ins/+	ins/+	ins/+	1	
13	106781964	106782734	Insertion	ins/+	ins/+	ins/+	1	
13	108681325	108692408	Insertion	ins/+	ins/+	ins/+	1	
13	112258885	112258885	Insertion	ins/+	del/ins	del/ins	1	
13	112275337	112327618	Insertion	ins1/ins2	ins2/+	ins1/+	1	
14	21269060	21280830	Insertion	ins/+	ins/ins*	+/+	1	
14	23971408	23973691	Insertion	ins/+	ins/+	ins/+	1	
14	30680034	30689300	Insertion	+/+	ins/+	ins/+	1	
14	39519582	39526044	Insertion	+/+	+/+	ins/+	1	
14	39525659	39525659	Insertion	+/+	+/+	ins/+	1	
14	46350239	46375297	Insertion	ins/+	ins/ins#	ins/+	1	
14	49753405	49753405	Insertion	ins/ins	ins/ins	ins/ins	1	
14	49753405	49784603	Insertion	ins/ins	ins/ins	ins/ins	1	
14	52186061	52204420	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
14	58760037	58760037	Insertion	ins/+	+/+	ins/+	1	
14	58760037	58767580	Insertion	ins/+	+/+	ins/+	1	
14	63795415	63806553	Insertion	ins/ins	ins/ins	ins/+	1	
14	66947122	66960697	Insertion	+/+	+/+	ins/+	1	
14	69027770	69034325	Insertion	ins/+	ins/+	ins/+	1	
14	85908492	85927961	Insertion	+/+	+/+	ins/+	1	
14	91631155	91640037	Insertion	ins/+#	ins/+	+/+#	1	
14	91631155	91631155	Insertion	ins/ins	ins/ins	ins/ins	1	
14	104297814	104304131	Insertion	+/+	ins/+	ins/+	1	
14	104496071	104499324	Insertion	ins/ins	ins/ins	ins/ins*	1	
14	104805271	104842647	Insertion	ins/+#	ins/+#	ins/+#	1	
14	105218672	105218672	Insertion	ins1/+	ins1/+	ins2/+	1	
14	105226516	105262417	Insertion	+/+	ins/+	ins/+	1	
14	106256448	106264936	Insertion	+/+	ins/+	+/+	1	
14	106256739	106264936	Insertion	+/+	ins/+	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
14	106745274	106759765	Insertion	+/+	ins/+	+/+	1	
15	20035650	20120289	Insertion	ins/+	ins/+	+/+	1	
15	20090577	20120289	Insertion	ins/+#	ins/+#	ins/+#	1	
15	20335244	20348914	Insertion	ins/+	ins/+	ins/+	1	
15	20335244	20357048	Insertion	ins/+	ins/+*	ins/+	1	
15	28404963	28506689	Insertion	ins/+	ins/+	ins/+	1	
15	29550872	29561055	Insertion	ins/ins	ins/ins	ins/ins	1	
15	33739612	33739612	Insertion	ins/ins	ins/+	ins/+	1	
15	33739620	33745698	Insertion	ins/ins	ins/+	ins/+	1	DGV
15	33739620	33745711	Insertion	ins/ins	ins/+	ins/+	1	DGV
15	34374148	34378163	Insertion	ins/ins	ins/ins	ins/ins	1	
15	47215114	47221200	Insertion	del/+*	ins/+	ins/+	0	DGV
15	47215114	47221203	Insertion	del/+*	ins/+	ins/+	0	DGV
15	55945681	55959074	Insertion	ins/+	+/+	ins/+	1	
15	55946064	55959074	Insertion	ins/+	+/+	ins/+	1	
15	66101187	66101187	Insertion	ins/+*	ins/+*	ins/+*	1	
15	66101187	66108397	Insertion	ins/+*	ins/+*	ins/+*	1	
15	66109459	66119524	Insertion	ins/+#	ins/+#	+/+#	1	
15	68390638	68391387	Insertion	ins/ins	ins/ins	ins/+	1	
15	68391012	68401959	Insertion	ins/ins	ins/ins	ins/+	1	
15	77028085	77067517	Insertion	+/+	+/+	ins/+	1	
15	77689302	77702759	Insertion	ins/ins	ins/ins	ins/ins	1	
15	83225654	83247745	Insertion	+/+	ins/+	ins/+#	1	
15	100705854	100715174	Insertion	ins/ins	ins/ins	ins/ins	1	
15	101530365	101533657	Insertion	ins/+	ins/ins	+/+	1	
15	101537059	101547744	Insertion	ins/+	+/+	ins/ins	1	
15	101537059	101549949	Insertion	ins/+	+/+	ins/ins	1	
15	101747832	101769186	Insertion	ins1/+	ins1/ins2	ins2/+	1	
15	101778928	101780127	Insertion	ins/+	ins/+	+/+	1	
16	845823	910898	Insertion	ins/ins	ins/ins#	ins/ins#	1	
16	1022828	1025835	Insertion	ins/ins*	ins/ins	ins/+	1	
16	1171855	1186785	Insertion	ins/+	+/+*	ins/+	1	
16	1237243	1249198	Insertion	+/+	+/+	ins/+	1	
16	1242880	1249198	Insertion	+/+	+/+	ins/+	1	
16	2860675	2866455	Insertion	ins/ins	ins/ins	ins/ins	1	
16	12074187	12092053	Insertion	+/+	ins/+*	ins/+	1	
16	15105499	15135097	Insertion	ins/+*	ins/+	NC	1	
16	18319179	18359332	Insertion	ins/+	+/+	ins/+*	1	
16	21209296	21223018	Insertion	ins/+	ins/ins#	ins/+	1	
16	24848111	24892299	Insertion	ins/+*	ins/ins	ins/+#	1	
16	26171275	26179244	Insertion	ins/ins	ins/ins	ins/+	1	
16	32271993	32306977	Insertion	ins1/ins2	ins1/ins2	ins1/ins2	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
16	33478024	33513241	Insertion	ins/+#	ins/+#	ins/+#	1	
16	65127339	65130323	Insertion	ins/ins	ins/ins	ins/ins	1	
16	69183165	69193804	Insertion	ins/ins	ins/ins	ins/ins	1	
16	69193804	69193804	Insertion	ins/ins	ins/ins	ins/ins	1	
16	69193804	69199372	Insertion	ins/ins	ins/ins	ins/ins	1	
16	69199372	69219306	Insertion	ins/ins	ins/ins	ins/ins	1	
16	69955864	69962532	Insertion	ins/+	ins/+	ins/+	1	
16	69962532	69962532	Insertion	ins/+	ins/+	ins/+	1	
16	69962532	69967982	Insertion	ins/+	ins/+	ins/+	1	
16	71283414	71302481	Insertion	ins/ins*	ins/ins*	ins/+	1	
16	76005413	76034042	Insertion	ins/ins	ins/ins	ins/ins	1	
16	76005413	76034478	Insertion	ins/ins	ins/ins	ins/+	1	
16	86985730	86988880	Insertion	ins/ins	ins/ins	ins/ins	1	
16	88730120	88734493	Insertion	ins/ins	ins/ins	ins/ins	1	
16	89625047	89649524	Insertion	ins/+	+/+	ins/+	1	
17	284873	319133	Insertion	ins/ins*	ins/ins*	ins/ins	1	
17	293106	319133	Insertion	ins/+*	ins/+*	ins/ins	1	
17	936331	976627	Insertion	ins/ins	ins/+	ins/+	1	
17	937312	976627	Insertion	ins/ins	ins/+	ins/+	1	
17	1057159	1067408	Insertion	ins/+*	ins/+#	ins/+*	1	
17	1300220	1314916	Insertion	+/+	ins/+	+/+	1	
17	6191740	6203596	Insertion	ins1/ins2	ins1/ins2	ins1/ins1	1	
17	7291925	7310537	Insertion	ins/ins	ins/+	ins/+	1	
17	8401866	8410909	Insertion	ins/+	ins/ins	+/+	1	
17	16824006	16824006	Insertion	+/+	+/+	ins/+	1	
17	18362831	18379209	Insertion	ins/+	ins/+	ins/+	1	
17	22375007	22412371	Insertion	ins/+#	ins/+#	ins/+#	1	
17	36441590	36454803	Insertion	+/+	ins/+	+/+	1	
17	41629109	41637071	Insertion	ins/+	ins/+	ins/+*	1	
17	43229876	43356253	Insertion	ins1/ins2*	ins1/ins2*	ins2/ins2*	1	
17	45511762	45514509	Insertion	ins/+#	ins/+#	ins/+#	1	
17	45514509	45518502	Insertion	ins/+	ins/+	ins/+	1	
17	45533148	45533148	Insertion	ins/ins	ins/+*	ins/ins	1	
17	46274712	46453424	Insertion	ins/+	ins/+	ins/+	1	
17	46274712	46294527	Insertion	+/+	ins/+	+/+	1	
17	46337328	46453424	Insertion	NC	ins/+	+/+	NC	
17	50037691	50045735	Insertion	ins/ins	ins/ins	ins/ins	1	
17	52582403	52611216	Insertion	ins/ins*	ins/+	ins/ins	1	
17	66623846	66636867	Insertion	ins/+	ins/+	+/+	1	
17	66636867	66636867	Insertion	ins/+	ins/+	+/+	1	
17	70361240	70367289	Insertion	ins/+	+/+	ins/ins	1	DGV
17	70361240	70367317	Insertion	ins/+	+/+	ins/ins	1	DGV

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
17	72829481	72884868	Insertion	ins/ins	ins/ins	ins/+	1	
17	72829481	72885525	Insertion	ins1/ins1	ins1/ins1	ins1/ins2	1	
17	72829481	72886182	Insertion	+/+	+/+	ins/+	1	
17	76067233	76074973	Insertion	ins/ins	ins/ins	ins/ins*	1	
17	76321691	76326980	Insertion	+/+	+/+	ins/+	1	
17	81241578	81265592	Insertion	+/+	ins/+	+/+	1	
17	81336003	81355216	Insertion	ins/ins	ins/+	ins/+	1	
17	82574686	82592645	Insertion	ins/ins	ins/ins	ins/ins*	1	
18	1833533	1851301	Insertion	ins/+	ins/+	+/+	1	
18	3100813	3112487	Insertion	ins/ins	ins/ins	ins/ins	1	
18	12484407	12485664	Insertion	ins/+	+/+	ins/+	1	
18	12485035	12485035	Insertion	ins/+	+/+	ins/+	1	
18	15078493	15104297	Insertion	ins/ins*	ins/+	ins/+	1	
18	22550622	22552873	Insertion	ins/ins	ins/ins	ins/ins	1	
18	43729360	43741177	Insertion	ins/ins	ins/+	ins/ins	1	
18	43741177	43741177	Insertion	ins/ins	ins/+	ins/ins	1	
18	48664671	48673399	Insertion	ins/+	ins/ins	ins/+	1	
18	50442045	50449944	Insertion	ins/+	ins/+	+/+	1	
18	53893756	53897171	Insertion	+/+	+/+	ins/+	1	
18	53899338	53905416	Insertion	+/+	+/+	ins/+	1	DGV
18	67452712	67456905	Insertion	ins/ins	ins/ins	ins/ins	1	
18	70461672	70469325	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
18	70474213	70474213	Insertion	ins/ins	ins/ins	ins/ins*	1	
18	72971017	72973672	Insertion	ins/+	ins/+	ins/+	1	
18	74199276	74212314	Insertion	ins1/ins1*	ins1/ins2	ins1/ins1*	1	
18	78229382	78235384	Insertion	ins/ins	ins/ins	ins/ins	1	
18	78235384	78239216	Insertion	ins/ins	ins/ins	ins/ins	1	
18	78238530	78238530	Insertion	ins/ins	ins/ins	ins/ins	1	
18	78487691	78521652	Insertion	ins/ins	NC	NC	NC	
18	79031233	79060989	Insertion	ins/ins*	ins/ins	ins/ins	1	
18	79611125	79626034	Insertion	ins/+	ins/ins	+/+	1	
19	125084	149226	Insertion	ins/+	ins/+	ins/+	1	
19	326559	434633	Insertion	ins/+	ins/+	ins/ins*	1	
19	860797	875441	Insertion	ins/ins	ins/ins	ins/ins	1	
19	862778	875441	Insertion	ins/ins	ins/ins	ins/ins	1	
19	875441	883322	Insertion	ins/ins	ins/ins	ins/ins	1	
19	1046457	1058044	Insertion	ins/+*	ins/ins	ins/+	1	
19	1136571	1154910	Insertion	ins/ins*	ins/ins	ins/ins*	1	
19	1159892	1170343	Insertion	ins/+*	ins/ins*	ins/+*	1	
19	3987779	4032053	Insertion	ins/ins*	ins/ins*	ins/+*	1	
19	3987779	4042755	Insertion	ins/ins*	ins/ins*	ins/+*	1	
19	6990508	7063542	Insertion	ins/+	ins/ins	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
19	7011552	7063542	Insertion	+/+	ins/+	+/+	1	
19	7441859	7452411	Insertion	ins1/ins2	ins1/ins1	NC	NC	
19	8775037	8780482	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
19	8879328	8889149	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
19	8895315	8898244	Insertion	ins/+*	ins/+*	ins/+*	1	
19	20399325	20410456	Insertion	ins/ins	ins/ins	ins/ins	1	
19	20809351	20826951	Insertion	ins/+	ins/+	+/+	1	
19	20826951	20826951	Insertion	ins/+	ins/+	+/+	1	
19	21572190	21591001	Insertion	ins/+	ins/+*	ins/+	1	
19	21591001	21591001	Insertion	ins/+	ins/+	ins/+	1	
19	22225726	22240218	Insertion	+/+	+/+	ins/+	1	
19	22763320	22779789	Insertion	ins/ins#	ins/ins#	NC	NC	
19	23827798	23866040	Insertion	ins/ins	ins/ins	ins/ins	1	
19	23843552	23866040	Insertion	ins/ins#	ins/ins*	ins/ins	1	
19	34233874	34248829	Insertion	ins/ins	ins/ins	ins/ins	1	
19	34385975	34393027	Insertion	ins/ins	ins/ins	ins/ins	1	
19	34409373	34411456	Insertion	ins/ins	ins/ins	ins/ins	1	
19	36184211	36233827	Insertion	ins/+*	+/+	ins/ins*	1	
19	37274336	37276874	Insertion	NC	ins/+	NC	NC	
19	39882578	39898829	Insertion	ins/+	ins/+	ins/+	1	
19	43315819	43329852	Insertion	ins/+	ins/ins	ins/+	1	
19	47416951	47420964	Insertion	ins/ins	ins/ins	ins/ins	1	
19	47853326	47959374	Insertion	ins/+*	ins/ins	ins/+*	1	
19	49163485	49185100	Insertion	ins/+	ins/+	ins/ins*	1	
19	49453259	49465756	Insertion	ins/+	ins/+	ins/+	1	
19	50066325	50144110	Insertion	ins/ins#	ins/ins*	ins/ins#	1	
19	54133553	54139458	Insertion	ins/ins*	ins/ins	ins/ins*	1	
19	54133553	54139829	Insertion	ins/+	ins/ins	NC	NC	
19	54234423	54297151	Insertion	ins/+	ins/+	ins/+	1	
19	54736273	54752668	Insertion	ins/+*	ins/+*	ins/ins*	1	
19	54752668	54848203	Insertion	ins/+	ins/+	ins/ins*	1	
19	54798596	54848203	Insertion	ins/+	+/+	ins/ins	1	
19	55712751	55722571	Insertion	ins/ins	ins/ins	ins/+	1	
20	1298060	1306346	Insertion	+/+	+/+	ins/+	1	
20	1875730	1878618	Insertion	ins/+	ins/ins	ins/+	1	
20	1875730	1890907	Insertion	ins/+	ins/ins	ins/+	1	
20	4093435	4136402	Insertion	ins/+*	ins/+*	ins/+*	1	
20	4440765	4450909	Insertion	ins/+	ins/+	ins/+	1	
20	5720368	5745967	Insertion	ins/+	ins/ins	ins/+	1	
20	13947646	13947646	Insertion	ins/ins	ins/ins	ins/ins	1	
20	13947646	13961181	Insertion	ins/ins*	ins/ins*	ins/+*	1	
20	38422110	38426307	Insertion	ins/+	ins/+	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
20	43754944	43766506	Insertion	+/+	ins/+	+/+	1	
20	47832962	47845840	Insertion	ins/+	ins/ins	+/+	1	
20	47832962	47898062	Insertion	ins/ins	ins/ins*	ins/ins*	1	
20	47888166	47898424	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
20	49613723	49618723	Insertion	+/+	ins/+	+/+	1	
20	62312810	62319804	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
20	63166978	63169767	Insertion	ins/+	ins/+	ins/+	1	
20	63692843	63697457	Insertion	ins/+	ins/+	ins/+	1	
20	63692843	63708831	Insertion	ins/+	ins/+	ins/+	1	
20	64084092	64106550	Insertion	ins/ins	ins/ins*	ins/ins*	1	
21	8372178	8387941	Insertion	ins/+	ins/+	ins/+	1	
21	8372178	8389456	Insertion	ins/+	ins/+	ins/+	1	
21	9045264	9064392	Insertion	ins/+	ins/+	ins/+	1	
21	9064392	9071385	Insertion	+/+	ins/+	+/+	1	
21	9807738	9837956	Insertion	ins/+	+/+	ins/+	1	
21	9810095	9837956	Insertion	ins/+	ins/+	ins/+	1	
21	14439191	14443151	Insertion	ins/ins	ins/ins	ins/ins	1	
21	17713489	17740638	Insertion	ins/ins	ins/ins	ins/ins	1	
21	18131823	18153524	Insertion	ins/ins	ins/+*	ins/+*	1	
21	22243007	22244486	Insertion	+/+	+/+	ins/+	1	
21	29116755	29123795	Insertion	ins/ins	ins/ins	ins/ins	1	
21	33317990	33324542	Insertion	ins/ins	ins/ins	ins/ins	1	
21	33463348	33533959	Insertion	ins/+	ins/+	ins/+*	1	
21	39970216	39976188	Insertion	ins/+	ins/+	ins/ins	1	
21	44405187	44414027	Insertion	ins/ins	ins/ins	ins/ins*	1	
21	44418179	44428421	Insertion	ins/ins	ins/+*	ins/+	1	
21	45796360	45801495	Insertion	ins/ins	ins/ins	ins/ins	1	
21	46287993	46299112	Insertion	ins1/ins2	ins1/ins1	ins2/ins2	1	
21	46287993	46299601	Insertion	ins/ins	ins/ins	ins/ins	1	
22	15927986	15983810	Insertion	ins/+	ins/+	ins/ins*	1	
22	15939531	15983810	Insertion	ins/+#	+/+	ins/ins#	1	
22	16767628	16768398	Insertion	+/+	ins/+	+/+	1	
22	17873372	17882208	Insertion	ins1/ins2	ins1/ins2	ins1/ins1	1	
22	19217826	19218760	Insertion	+/+	+/+	ins/+	1	
22	19223337	19229410	Insertion	+/+	+/+	ins/+	1	DGV
22	20331920	20352195	Insertion	ins/ins	ins/ins*	ins/+	1	
22	20331920	20383088	Insertion	+/+	+/+	ins/+	1	
22	20489693	20508324	Insertion	ins/ins	ins/ins	ins/ins	1	
22	21311654	21332751	Insertion	ins/+*	ins/+*	ins/+*	1	
22	22353689	22357050	Insertion	+/+	ins/+	ins/+	1	
22	22361901	22363316	Insertion	+/+	ins/+	ins/+	1	
22	22902064	22907724	Insertion	+/+	ins/+	+/+	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
22	22907724	22907724	Insertion	+/+	ins/+	+/+	1	
22	23510121	23519375	Insertion	ins/ins	ins/ins	ins/ins	1	
22	23928615	24006189	Insertion	ins/+	ins/+	+/+	1	
22	23929540	23945653	Insertion	ins/ins	ins/ins#	ins/+#	1	
22	23969194	23989011	Insertion	ins/+	ins/ins*	+/+	1	
22	35726767	35731686	Insertion	ins/ins	ins/+	ins/+	1	
22	35731686	35745486	Insertion	ins/ins	ins/+	ins/+	1	
22	38643581	38651120	Insertion	+/+	+/+	ins/+	1	
22	41296876	41301512	Insertion	ins/ins	ins/ins	ins/+	1	
22	42128704	42134001	Insertion	ins/+	ins/+	+/+	1	
22	42128704	42134634	Insertion	ins/+	ins/+	+/+	1	
22	45326000	45333075	Insertion	+/+	ins/+	+/+	1	
22	46469325	46472467	Insertion	ins/ins	ins/ins*	ins/ins*	1	
22	48977584	48991798	Insertion	ins/+	ins/+	ins/+	1	
22	48978217	48990253	Insertion	ins/ins	ins/ins	ins/+	1	
22	50641792	50655551	Insertion	ins/ins*	ins/ins	ins/ins*	1	
23	2311692	2324586	Insertion	ins/ins	ins/ins	ins/ins#	1	
23	2315797	2324586	Insertion	ins/ins	ins/ins	ins/ins#	1	
23	5043166	5052802	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
23	6514301	6559262	Insertion	ins/+	ins/+*	ins/+	1	
23	6514696	6559262	Insertion	ins/+	ins/+#	ins/+	1	
23	8154125	8187175	Insertion	ins/ins	ins/ins	ins/+	1	
23	9388660	9410729	Insertion	ins/+	ins/ins	ins/+	1	
23	9410729	9421447	Insertion	ins/+	+/+	ins/+	1	
23	26778253	26788141	Insertion	ins/+	+/+	ins/+	1	
23	30771481	30779411	Insertion	ins/+#	ins/ins#	+/+	1	
23	47087595	47087595	Insertion	ins/+	+/+	ins/+	1	
23	50053230	50061678	Insertion	ins/ins	ins/ins	ins/ins	1	
23	52074439	52085788	Insertion	+/+	+/+	ins/+	1	
23	52500704	52508634	Insertion	ins/+*	+/+	ins/+*	1	
23	52509230	52509230	Insertion	ins/ins*	ins/ins*	ins/+	1	
23	52509827	52512179	Insertion	ins/+	+/+	ins/+	1	
23	52521151	52534018	Insertion	ins/+	ins/ins	ins/+	1	
23	55644794	55653716	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
23	56764920	56770925	Insertion	ins/+	ins/ins	ins/+	1	
23	56770925	56776327	Insertion	ins/+	ins/ins	ins/+	1	
23	56776327	56776327	Insertion	ins/+	ins/+	+/+	1	
23	56776327	56784945	Insertion	ins/+	ins/ins	ins/+	1	
23	62963266	62975334	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
23	89199776	89223644	Insertion	ins/ins	ins/ins	ins/+	1	
23	90260365	90262703	Insertion	ins/ins	NC	ins/ins	NC	
23	90262703	90277618	Insertion	ins/+*	ins/+	ins/ins	1	

chr	start	stop	sv	NA12878	NA12891	NA12892	Mendelian	Method
23	101810173	101811805	Insertion	ins/+	ins/ins#	+/+	1	
23	101811805	101811805	Insertion	ins/+	ins/ins	NC	NC	
23	108762221	108762221	Insertion	ins/ins	ins/ins#	ins/ins#	1	
23	108771648	108795396	Insertion	ins/+	ins/+	ins/ins*	1	
23	108772220	108795396	Insertion	ins/ins#	ins/ins#	ins/ins#	1	
23	108773126	108795396	Insertion	ins/ins	ins/ins#	ins/ins#	1	
23	112297176	112312651	Insertion	ins/+	ins/ins	+/+	1	
23	112312651	112312651	Insertion	ins/+	ins/ins	+/+	1	
23	112314505	112320591	Insertion	ins/+	ins/ins	+/+	1	DGV
23	112597654	112609925	Insertion	ins/+	ins/ins	+/+	1	
23	112598072	112609925	Insertion	ins/+	ins/ins	+/+	1	
23	116010268	116044138	Insertion	ins/ins*	ins/+*	ins/+*	1	
23	116010268	116044578	Insertion	ins/ins#	ins/+#	ins/+#	1	
23	128466230	128471036	Insertion	ins/ins	ins/ins	ins/ins	1	
23	128466786	128471036	Insertion	ins/ins	ins/ins	ins/ins	1	
23	131129210	131156754	Insertion	ins/ins	ins/ins#	ins/ins	1	
23	135719353	135727553	Insertion	ins/+	+/+	ins/+	1	
23	139494667	139525975	Insertion	ins/ins*	ins/ins*	ins/ins*	1	
23	140716910	140727165	Insertion	ins/ins	ins/ins*	ins/ins*	1	
23	140716910	140727686	Insertion	ins/ins	ins/ins	ins/ins	1	
23	140991769	140995989	Insertion	ins/+	+/+	ins/+	1	
23	141007688	141018366	Insertion	ins/+	+/+	ins/+	1	
23	147269450	147298842	Insertion	ins/ins	ins/ins	ins/ins*	1	
23	153378518	153398095	Insertion	ins/+	ins/+*	ins/ins*	1	
23	154642708	154655010	Insertion	ins/+	ins/ins	+/+	1	
24	7708840	7723973	Insertion	NC	ins/ins	NC	NC	
24	9907429	9913864	Insertion	NC	ins/+	NC	NC	
24	9913864	9917945	Insertion	NC	ins/+	NC	NC	

Table S3 Validation of deletion calls by publicly available nextr generation sequencing (NGS) data.

	NA12878	NA12891	NA12892
No. of deletion investigated*	503	475	438
Consistent with NGS	464	426	372
Inconsistent with NGS	39	49	66
Percentage consistent with NGS	92%	90%	85%

* NGS validation was only applied to loci that could be mapped to hg19.

Table S4 Inversion detected in NA12878.

Chr	Start	Stop	Notes
1	16890650	17181198	novel
2	97199286	97581211	Kidd *et al.* 2008
2	130857096	132332612	novel
6	26673441	26871078	Kidd *et al.* 2008
7	54199302	54342749	Kidd *et al.* 2008
7	54300348	54377440	novel
7	143910299	143983189	novel
7	144160778	144412427	Kidd *et al.* 2008
7	149756935	153761206	novel
7	149756935	153761206	novel
8	2186318	2291582	novel
8	2244521	2475337	Kidd *et al.* 2008
8	2304096	2516024	Kidd *et al.* 2008
10	24492511	24521895	Kidd *et al.* 2008
11	4242987	4344342	Kidd *et al.* 2008
11	50131500	50421805	Kidd *et al.* 2008
11	89811778	90079788	Kidd *et al.* 2008
12	17739217	17899533	Kidd *et al.* 2008
14	19350902	20195876	novel
16	2536472	2680329	Kidd *et al.* 2008
16	14784574	15409607	novel
16	14906944	15409607	novel
16	21417358	22712952	novel
23	52661863	52825007	Kidd *et al.* 2008
23	71684267	71835832	Kidd *et al.* 2008
23	103940531	104117650	Kidd *et al.* 2008
23	120006842	120219579	Kidd *et al.* 2008

File S1 Supporting Table S1 and S2 legends.

Legends for Table S1

Genotype call

ins	insertion allele; ins1, ins2, …, indicates insertion alleles of different sizes.
del	deletion allele; del1, del2, …, indicates deletion alleles of different sizes.
+	reference allele
NC	no genotype calls were made
#	low confidence genotype call due to <50X depth of coverage
*	low conference call on zygosity (het vs. hom)

Notes

DGV	Validated published SV

Legends for Table S2

Genotype call

ins	insertion allele; ins1, ins2, …, indicates insertion alleles of different sizes.
del	deletion allele; del1, del2, …, indicates deletion alleles of different sizes.
+	reference allele
NC	no genotype calls were made
#	low confidence genotype call due to <50X depth of coverage
*	low conference call on zygosity (het vs. hom)

Notes

DGV	validated published SV

cnp / spratio

cnp	coverage depth method
spratio	s/(s+p) ratios method
1	evidence of deletion
0	no evidence of deletion
[blank]	hg38 regions unmappable to hg19 coordinates for NGS validation