

ONLINE METHODS

Study design and microarrays. Six samples (NA10851, NA12239, NA18517, NA18576, NA18980 and NA15510) were run on all eleven platforms in triplicate. Experiments on higher resolution platforms were independently performed in two different laboratories, whereas lower resolution arrays were run at one site (summarized in **Table 1**), and data analysis for each algorithm performed at one site. For all CGH platforms sample NA10851 was used as reference DNA in competitive hybridization experiments. For these platforms, one set of triplicates represented self-self hybridizations with the NA10851 sample. Samples were required to pass initial platform-specific quality control, as well as additional CNV-specific quality control filters that were systematically applied to all CNV data sets generated by each algorithm (**Supplementary Methods**). A total of eleven CNV calling algorithms were used, where each array data set was analyzed with one to six distinct tools (**Table 1**). Description of each array platform and CNV methods can be found in **Supplementary Methods**, including description of the parameters and settings used for each algorithm. A minimum of five probes and 1 kb length were required for all CNV calls, with the exception

of the BAC array for which calls from a single clone was allowed. A list of all CNVs passing quality control is available in **Supplementary Table 3**.

Reference 'gold standard' data sets. To evaluate CNV calling reproducibility, five different data sets were prepared: (i) a high-resolution array-based subset of the Database of Genomic Variants (DGV), (ii) a sequencing-based DGV subset, (iii) an ultra-high resolution aCGH-based set of 8,599 validated CNVs¹¹, (iv) a CNV genotype data for 4,978 variants¹¹ and (v) 1,157 deletions derived from paired-end mapping based on fosmid-end sequencing⁴¹. To evaluate breakpoint precision, two nucleotide-resolution breakpoint data sets were used: (i) a set of 862 nonredundant deletion breakpoints compiled from two published sources, a library of breakpoints collated from personal sequencing projects⁴⁴ and a set of breakpoints derived from targeted hybridization-based DNA capture and 454 sequencing of all array-based CNVs detected in three unrelated individuals⁴³, and (ii) a set of sample-level deletion breakpoints derived from four samples sequenced in the 1000 Genomes Project^{45,46}. A detailed description of these analyses and the parameters used to define overlap and accuracy is provided in **Supplementary Methods**.