karyotype are presented as draggable objects and can be moved by mouse or by gestures when using a touch screen. To complete a karyotype the chromosomes have to be moved to the correct position of a karyogram form. The students submit the completed karyogram to the system. The karyogram is rejected if any chromosome has not been placed at its correct position, and the students have to correct the mistake before they resubmit the karyogram. If the karyogram has been accepted, the students have to write the correct karyotype. The karyotype is rejected if it is not written according to ISCN. A course containing seven different karyotypes including numerical as well as structural chromosomal changes was developed and was integrated in the learning management system ILIAS as a sharable content object reference (SCORM) module. In a practical lesson, which was attended by 150 students, each student accessed the course using his own mobile electronic device via WIFI. The students were assisted by cytogeneticists so that each student was able to complete at least one online karyotype. Thus, the online karyotyping course proved to be a robust and platform independent tool for teaching cytogenetics. The system may also be used to address different levels of cytogenetic skills and/or specific learning objectives such as complex karyotypes in acute leukemias, for instance, in graduate medical education and cytogenetic specialization. Supported by the eLearning Foederfonds of the Heinrich-Heine-University, Duesseldorf

6. Genomics

6.P1

Inversion variants in human and primate genomes

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For many years, inversions have been proposed to be a direct driving force in speciation since they suppress recombination when heterozygous. Inversions are the most common large-scale differences among humans and great apes. Nevertheless, they represent large events easily distinguishable by classical cytogenetics, whose resolution, however, is limited. Here, we performed a genome-wide comparison between human, great ape, and macaque genomes using the net alignments for the most recent releases of genome assemblies. We identified a total of 156 putative inversions, between 103 kb and 91 Mb, cor- responding to 136 human loci. Combining literature, sequence, and experimental analyses, we analyzed 109 of these loci and found 67 regions inverted in one or multiple primates, including 28 newly identified inversions. These events overlap with 81 human genes at their breakpoints, and seven correspond to sites of recurrent rearrangements associated with human dis- ease. This work doubles the number of validated primate inversions larger than 100 kb, beyond what was previously doc- umented. We identified 74 sites of errors, where the sequence has been assembled in the wrong orientation, in the reference genomes analyzed. Our data serve two purposes: First, we generated a map of evolutionary inversions in these genomes representing a resource for interrogating differences among these species at a functional level; second, we provide a list of misassembled regions in these primate genomes, involving over 300 Mb of DNA and 1978 human genes. Accurately annotating these regions in the genome references has immediate applications for evolutionary and biomedical studies on primates.

6.P2

Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus

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Human chromosome 15q25 is involved in several disease-associated structural rearrangements, including microdeletions and chromosomal markers with inverted duplications. Using comparative fluorescence in situ hybridization, strand-sequencing, single-molecule, real-time sequencing and Bionano optical mapping analyses, we investigated the organization of the 15q25 region in human and nonhuman primates. We found that two independent inversions occurred in this region after the fission event that gave rise to phylogenetic chromosomes XIV and XV in humans and great apes. One of these inversions is still polymorphic in the human population today and may confer differential susceptibility to 15q25 microdeletions and inverted duplications. The inversion breakpoints map within segmental duplications containing core duplicons of the GOLGA gene family and correspond to the site of an ancestral centromere, which became inactivated about 25 million years ago. The inactivation of this centromere likely released segmental duplications from recombination repression typical of centromeric regions. This increased the frequency of ectopic recombination creating a hotspot of hominid inversions where dispersed GOLGA core elements now predispose this region to recurrent genomic rearrangements associated with disease.

6.P3

Characterization of structural variation of the human glycophorin locus using multicolour fibre FISH

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Structural variants (SVs) represent an important source of variation among individual human genomes. These variants are prone to arise in repetitive regions and may involve multiallelic gene families with internal complex structures, which make its characterization a challenging task. Multicolor fibre-FISH on combed DNA-fibres has been successfully used for the visualization and characterization of structural variants, leading to a better interpretation and understanding of genomic structural variation identified by different genomic technologies. The human glycophorin gene cluster is known to undergo extensive structural variation and gene conversion. This region harbours the tandemly arranged glycophorin genes GYPE, GYPB and GYPA, sharing ~97% identity. Glycophorin A and glycophorin B are red blood cell surface proteins and act as receptors for the parasite