GENETICS

FOR THE FIRST TIME TWO HUMAN GENOMES COMPARED

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Another “first discovery” in this nature genetics preprint although the analysis could have already been done some years earlier. The CNV specialists from Toronto now compare the Human Genome Project sequence with the Celera sequence – the gap between the two compilations was obviously bigger than the intra-sequence gaps. Of course both sequences are still mosaics from several individuals but the analysis nicely exemplifies how difficult it will be to compare the genome of two different human beings.
The authors employ a whole battery of alignment tools BLAT, MEGABLAST, GCA and A2Amapper. Of course results depend on the strategy, definition and implementation. As show by FISH analysis most of the discrepancies are true and can be classified into a few categories – insertions or deletions if seen from the second genome (has somebody ever thought about a minimal human genome?), mismatches and inversions. We are getting here a preview of the diagnostic workup in a patient in 2026. This blog contains forward looking statements while the responsibility rests solely with the reader. Yea, yea.