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Detecting copy number changes in genomic DNA - MAPH and MLPA

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STELLINGEN

Behorende bij het proefschrift

"Detecting copy number changes in genomic DNA – MAPH and MLPA"

1. Copy number polymorphisms are widespread throughout the genome and contribute to interindividual variation.
(this thesis)
Hollox *et al.*, 2002. *Am. J. Hum. Genet.* 73:591-600.
Iafrate *et al.*, 2004. *Nat. Genet.* 36: 949-951.
Sebat *et al.*, 2004. *Science.* 305:525-528.
2. Duplications and deletions within the *DMD* gene have a different distribution, and are probably caused by different mechanisms.
(this thesis)
3. Rubinstein-Taybi syndrome can be caused by mutations in either of the *CREBBP* or *EP300* genes.
(this thesis)
4. Genomic duplicons are complex structures that cannot be fully characterised by sequence analysis alone.
(this thesis)
She *et al.*, 2004. *Nature.* 431:927-930.
5. The term “junk DNA” is a classic example of assuming to be unimportant that which is not understood.
Mattick, 2004. *Nat. Rev. Genet.* 5:316-323.
Bejerano *et al.*, 2004. *Science.* 304:1321-1325.
6. That intragenic rearrangements may the cause of a given disease has been consistently underestimated in mutation screening.
7. The reason behind the relatively large size of the first intron of many genes deserves further investigation.

Wight *et al.*, 1997. *Gene.* 201:111-117
Pozzoli *et al.*, 2003. *Genome Res.* 13:764-772
8. It appears that the bovine bacillus of the *Mycobacterium tuberculosis* complex (*Mycobacterium bovis*) is derived from the human bacillus (*Mycobacterium tuberculosis*), not vice versa as was originally thought.

Cole *et al.*, 1998. *Nature.* 393:573-544
Garnier *et al.*, 2003. *Proc. Nat. Acad. Sci.* 100:7877-7882
9. Whoever said “if experiments always worked it would get boring” was obviously out of the lab for a long time.

10. The extra distance obtainable with modern golf clubs means for most golfers that the ball goes 20 meters further into the lake.
11. Leg-byes give an unfair advantage to the batting side and should be removed from cricket.
12. Reading Fokke and Sukke may not always give an accurate perspective on Dutch society, but it is an excellent way to expand your vocabulary.

Stefan White, februari 3, 2005.