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Advanced genome-wide screening in human genomic disorders

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Propositions

with the thesis

Advanced genome-wide screening in human genomic disorders

of

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- In a significant proportion of cases with a complex chromosomal translocation pattern classical G-banding only is not enough to elucidate the rearrangements. (*This thesis*)
- To elucidate the structure and complexity of a ring chromosome high resolution molecular investigations are always needed. (*This thesis*)
- Pooling normal copy number variation in a database such as the Database of Genomic Variants is of crucial importance for the correct interpretation of data, but it may also cause false negative calls in the case of polygenic involvement or homozygous deletions of the copy number variant. (*This thesis*)
- Together with a microduplication of 3q29 unknown genetic or epigenetic factors play a role in the determination of the phenotypic severity of this abnormality. (*This thesis*)
- As screening for genetic causes of idiopathic mental retardation remains a genome-wide job, increase of resolution will yield more genetic diagnoses but will also yield a burden of unwanted genetic information.
- Array-CGH will soon be replaced by sequencing techniques.
- Not the identification but the interpretation of copy number variation has become the challenge for the future.
- The term “normal” in normal copy number variation is misleading, since its influence will depend on the genetic background in several cases.
- Chromosome banding will remain indispensable for clinical cytogenetics to elucidate certain types of structural alterations.
- As in vitro cell culture systems are frequently instable, results obtained with commonly used cell lines should be interpreted with great care.
- Scientific publications that reach the general news bulletins are often interpreted incorrectly by the layman.
- Knowledge transfer is much more efficient at small and dedicated conferences than at large general congresses.
- Although almost all geographic maps imply that Belgium is situated under the Netherlands, this is not self-evident.