Karyotype is not dead (yet)!

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Résumé en anglais	BACKGROUND: While array-comparative genomic hybridization (a-CGH) and next-generation sequencing (NGS or exome) technologies have swiftly spread throughout the medical field, karyotype has gradually lost its leading role among genetic tests. Several international guidelines recommend starting with a-CGH screening then going on with exome analysis when investigating a patient with intellectual disability (ID) and no precise clinical diagnosis. A-CGH and whole exome sequencing increase etiologic diagnoses rate up to 30% in case of ID. However, physicians have to deal with the lack of qualitative information of the genome. Especially, exome and a-CGH analysis fail to detect chromosomal rearrangements because breakpoints are either located in introns or not associated with a gain or loss of genetic material. If these technologies cannot easily identify chromosomal translocations or inversions which sometimes split a gene, karyotype can. DISCUSSION: For the 5 cases described, karyotype provided the right diagnosis for a Mendelian disease while molecular analysis remained unsuccessful. We conclude that when a Mendelian disease is strongly suggested clinically, if molecular analysis is normal, it could be very useful to carry out a karyotype in order to demonstrate a chromosomal rearrangement involving the targeted gene. If this gene is disrupted, the physician can confirm the suspected disease and give appropriate genetic counseling. SUMMARY: This article aims at keeping in mind that karyotype, this old-fashioned genetic tool, can still remain powerful and useful within some genetic issues. Even in this modern period of whole exome sequencing, young geneticists should know that karyotype remains a powerful and cheap technology, available throughout the world and can still do a lot for families.

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