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Advantages of X-chromosomal microsatellites in deficiency paternity testing: presentation of cases

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Abstract. We present examples of our routine work, which demonstrate the impact of additional ChrX marker typing in special paternity case situations. In each of the seven cases, an extensive testing of autosomal markers failed to yield satisfying results (W >99.9% or 3 exclusions). However, by expanding the analysis to ChrX markers, these criterions were met in five of seven cases.

Keywords: ChrX marker typing; Deficiency paternity cases

1. Introduction

The major advantage of X-chromosomal (ChrX) STRs arises in deficiency paternity cases, i.e. when a putative father is not available and DNA from paternal relatives has to be analysed instead. In such cases, the exclusion power of autosomal STRs is substantially decreased, whereas ChrX markers are (at least in some cases) more efficient [1]. Female individuals fathered by the same man share their paternal ChrX. Males inherit their only ChrX from their mother. Hence, in cases in which the putative grandmother is available for genotyping, the possible ChrX alleles of the putative father can be determined [2]. ChrX marker typing is highly effective in mother–son kinship and in father–daughter testing. However, linkage and possible linkage disequilibrium between the ChrX markers used have to be taken into consideration.

2. Materials and methods

For the following pedigrees, we tested 11–16 autosomal STRs using the SGM or Powerplex 16 Kits and SE33, resulting in differing probabilities of paternity (W_{autos}).

Additionally, we investigated a panel of 9 ChrX STRs (see Fig. 1), but only four of it (the unlinked markers DXS6807, DXS8377 and the DXS101/DXS7424 haplotype frequency [3]) were used for calculation of probability of paternity (W_{ChrX}). The

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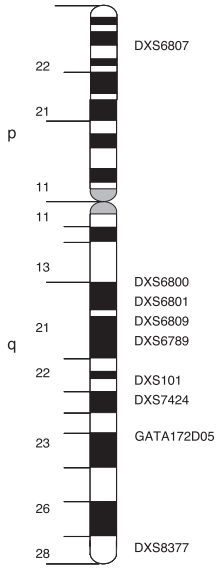
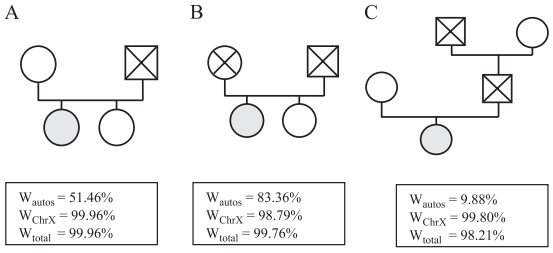


Fig. 1. ChrX ideogram with the investigated markers.

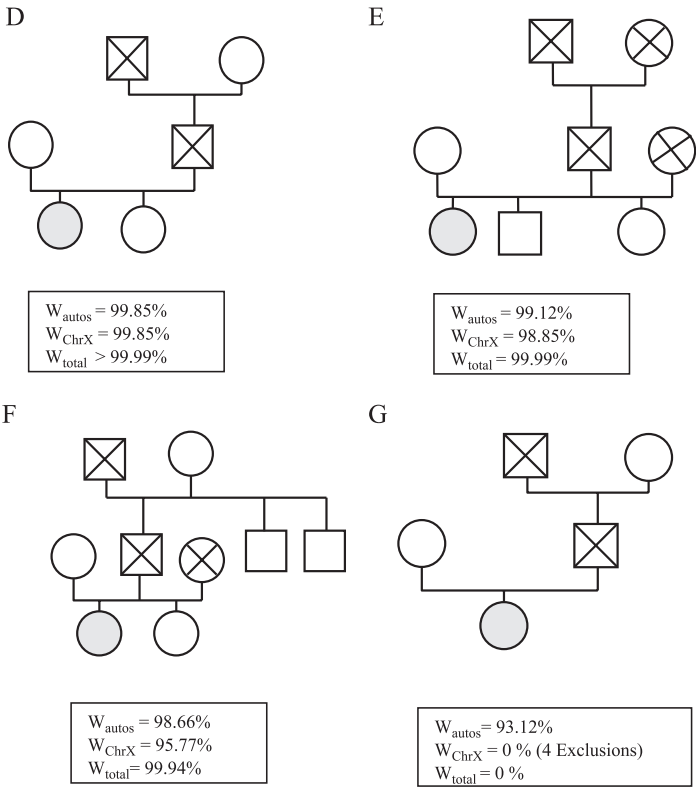
calculations for W_{autos} and W_{ChrX} were performed with the Kinship part of DNA-VIEW (Charles Brenner) considering the ChrX specific inheritance of the markers.

3. Results



4. Discussion

The presented examples of our routine work demonstrate the impact of information of an additional ChrX marker typing. In each of the cases, an extensive testing of autosomal markers failed to yield satisfying results (i.e. either W>99.9% or at least 3 exclusions). However, by expanding the analysis to ChrX markers, these criterions were met in five of seven cases. Two cases merit special attention: In case C, W_{autos} was 9.88% (‘‘paternity unlikely’’). ChrX analysis led to a W_{total} of 98.21%. In case G, W_{autos} was 93.12% when testing the putative grandmother. Including the brother of the putative father reduced the probability to 63.20% (not shown). However, in nine ChrX STRs, four exclusions were found.



These cases clearly demonstrate that ChrX markers have the potential to solve some paternity cases, which cannot be solved using autosomal markers. Suitable for this analysis are cases with a female child and especially putative grandmother-and putative sister cases. Female individuals with the same father share the same paternal ChrX. The ChrX marker investigation of two sisters or half-sisters can thus exclude paternity, even when none of the parents is available for testing. On the other hand, all ChrX alleles of the putative father can be determined by investigating the putative grandmother. However, ChrX marker genotypes of the putative grandmother can also be reconstructed from her children and if sons (brothers of the putative father) are available, the data are even more informative.

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