Diagnostic prénatale du Syndrome X Fragile
Prenatal diagnosis of Fragile X syndrome: small meiotic recombination events at the FMR1 locus

Abstract

**Objective** - Fragile X syndrome (FXS), the most common inherited cause of intellectual disability, is caused by an expansion over 200 CGG repeats (full mutation) in the FMR1 gene. Intergenerational instability of an expanded FMR1 allele is linked to the carrier's gender (female), the CGG repeat size and the number of AGG interspersions within the CGG repeat, making genetic counseling a complex task. The objective of our work was to emphasize the importance of combining haplotype analysis with FMR1 linked markers and CGG repeat sizing for prenatal diagnosis (PND) of FXS.

**Methods** - Two PND of FXS were performed using haplotype analyzes and sizing of the FMR1 allele.

**Results** - We detected two cases of meiotic recombination at the FMR1 locus, i.e., reciprocal double crossover or non-crossover, resulting in coexistence of the mutant maternal haplotype and the normal-size maternal CGG repeat.
Conclusion - These rare and unexpected cases (1/120 frequency in our experience) have to be kept in mind in PND of FXS since they prohibit using polymorphic marker haplotyping as the only tool to predict the fetus status.

This article is protected by copyright. All rights reserved.

#FXS #FMR1 #genetics #health

CONTACT :
Prof. Martine Raynaud : martine.raynaud@univ-tours.fr

À lire aussi

La régulation épigénétique de HsMar1, un transposon d'ADN humain

Intervention psychothérapeutique brève pour le traitement de la dépression d'intensité légère à modérée en soins primaires

Rencontre internationale Langage des Enfants avec Autisme

Haut de page