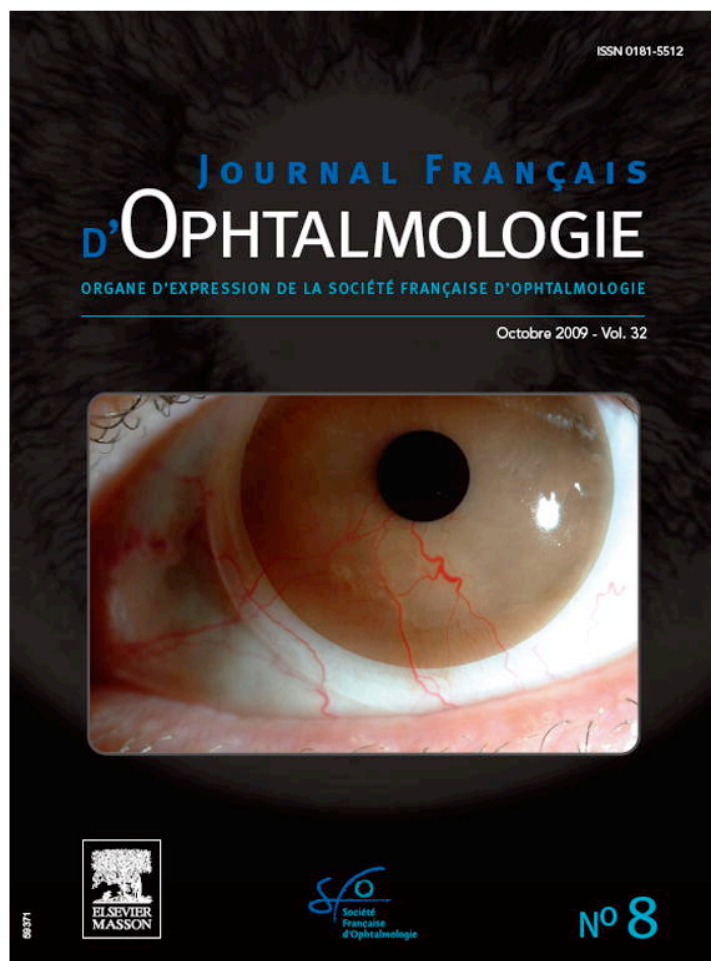


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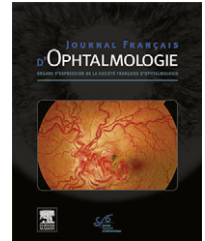
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ORIGINAL ARTICLE

Partial mosaic trisomy 5: A new case report with ocular involvement

Atteintes oculaires chez un enfant atteint d'une trisomie 5 partielle en mosaïque

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Summary We describe a new case of a male patient with a small marker chromosome present as 80% mosaic, derived from chromosome 5 with presence of posterior iridolenticular synechia, high hyperopia, epicanthic folds, hypertelorism, moderate developmental delay with lack of speech, macrocephaly, and subtle dysmorphic features including micrognathia, slightly rotated ears, and polydactyly. The karyotype of our patient was as follows: 46, XY/47, XY +mar, characterized by FISH (fluorescence in situ hybridization) using the chromosome five painting probe. Ocular involvement in trisomic 5 subjects is a very rare event. To our knowledge, only two cases have been described to date. The present case contributes to the description of the ocular presentation and the distinct clinical phenotype of *de novo* partial trisomy 5 syndrome. © 2009 Elsevier Masson SAS. All rights reserved.

Résumé Nous décrivons un nouveau cas de sexe masculin avec présence de mosaïcisme à 80% de petit marqueur dérivé du chromosome 5, présentant des synéchies postérieures iridolenticulaires, une hypermétropie forte, des replis épicanthiques avec hypertélorisme, un retard de développement modéré avec absence de langage, une macrocéphalie et quelques subtiles dysmorphies caractéristiques comme une micrognathie, les oreilles légèrement tournées et des doigts surnuméraires. Le caryotype du patient (46, XY/47, XY +mar) a été caractérisé par la méthode FISH (fluorescence in situ hybridization), en utilisant une sonde de coloration

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