

## Interesting case report ECARUCA (2013-10)

### Blepharophimosis, ptosis, epicanthus inversus syndrome with translocation and deletion at chromosome 3q23 in a black African female

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#### Abstract:

Blepharophimosis-epicanthus inversus syndrome (BPES) is a rare autosomal dominant disorder whose main features are the abnormal shape, position and alignment of the eyelids. Type I refers to BPES with female infertility from premature ovarian failure while type II is limited to the ocular features. A causative gene, FOXL2, has been localized to 3q23. We report a black female who carried a de novo chromosomal translocation and 3.13 Mb deletion at 3q23, 1.2 Mb 5' to FOXL2. This suggests the presence of distant cis regulatory elements at the extended FOXL2 locus. In spite of 21 protein coding genes in the 3.13 Mb deleted segment, the patient had no other malformation and a strictly normal psychomotor development at age 2.5 years. Our observation confirms panethnicity of BPES and adds to the knowledge of the complex cis regulation of human FOXL2 gene expression.

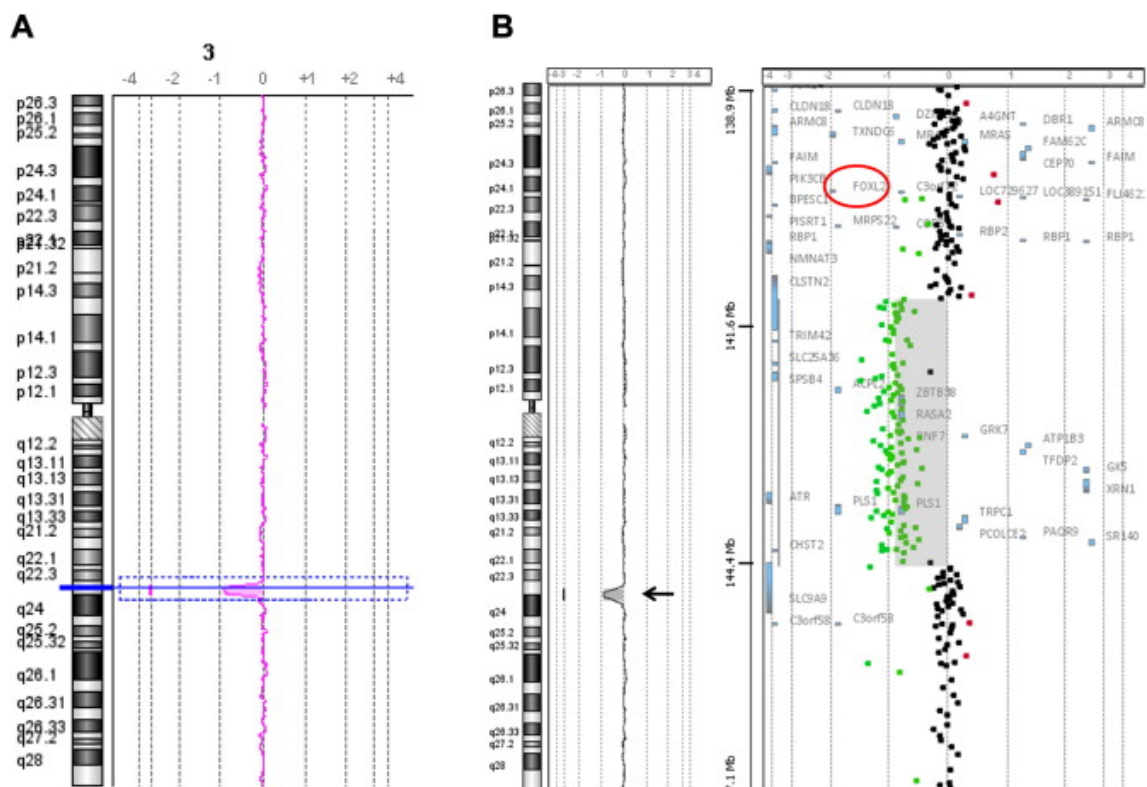


Fig. 1. Array-CGH (comparative genomic hybridization) analysis. A. Chromosomal view: the 3q23 deletion is indicated by the arrow. B. Gene view. Each dot represents an oligonucleotide probe. Blue boxes represent genes (FOXL2 is inside the red circle). The results are expressed as the log<sub>2</sub> of the ratio. A -0.7 to -1.2 ratio indicates that the oligonucleotide is deleted (green dots).

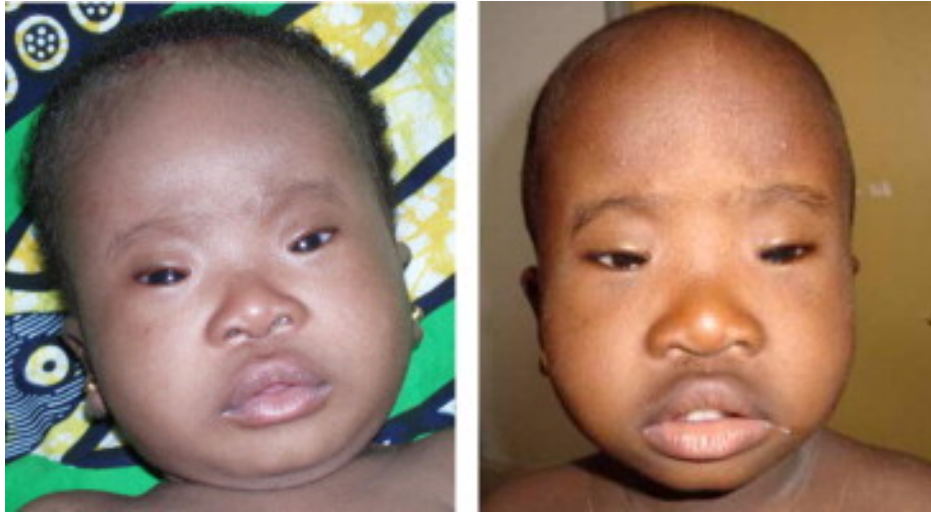


Fig. 2. Facial dysmorphism with telecanthus, blepharophimosis, upslanting palpebral fissures, flat nasal bridge and a relative macrostomia (at 3 and 12 months).