Get the latest research from NIH: https://www.nih.gov/coronavirus

The new PubMed site will become the default in mid-May. Click here to try it now! Frequently asked questions

Format: Abstract


**X-Linked Emery-Dreifuss Muscular Dystrophy: Study Of X-Chromosome Inactivation and Its Relation with Clinical Phenotypes in Female Carriers.**

Viggiano E¹, Madej-Pilarczyk A², Carboni N³, Picillo E¹, Ergoli M¹, Gaudio SD⁴, Marchel M⁵, Nigro G⁶, Palladino A¹, Politano L¹.

Author information

**Abstract**

X-linked Emery-Dreifuss muscular dystrophy (EDMD1) affects approximately 1:100,000 male births. Female carriers are usually asymptomatic but, in some cases, they may present clinical symptoms after age 50 at cardiac level, especially in the form of conduction tissue anomalies. The aim of this study was to evaluate the relation between heart involvement in symptomatic EDMD1 carriers and the X-chromosome inactivation (XCI) pattern. The XCI pattern was determined on the lymphocytes of 30 symptomatic and asymptomatic EDMD1 female carriers-25 familial and 5 sporadic cases-seeking genetic advice using the androgen receptor (AR) methylation-based assay. Carriers were subdivided according to whether they were above or below 50 years of age. A variance analysis was performed to compare the XCI pattern between symptomatic and asymptomatic carriers. The results show that 20% of EDMD1 carriers had cardiac symptoms, and that 50% of these were ≥50 years of age. The XCI pattern was similar in both symptomatic and asymptomatic carriers. Conclusions: Arrhythmias in EDMD1 carriers poorly correlate on lymphocytes to a skewed XCI, probably due to (a) the different embryological origin of cardiac conduction tissue compared to lymphocytes or (b) the preferential loss of atrial cells replaced by fibrous tissue.

**KEYWORDS**: Emery–Dreifuss muscular dystrophy (EDMD1); X-chromosome inactivation (XCI); cardiac symptoms; skewed X-chromosome inactivation

PMID: 31718017   PMCID: PMC6895991   DOI: 10.3390/genes10110919

[Indexed for MEDLINE]  Free PMC Article
