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# Genetic confirmation of germline mosaicism in a Duchenne muscular dystrophy Tunisian family

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**Introduction:** Duchenne muscular dystrophy (DMD) is a lethal neuromuscular disease. It is a dystrophinopathy with an X-linked recessive genetic disorder related to the absence of a cytoskeletal protein: The Dystrophin. In general, this disease affects predominantly only boys and women are vectors. In some cases, these women cannot have the gene defect responsible to the state of germline mosaicism. This situation was often suspected but seldom confirmed by genetic studies.

**Objective:** In this context, we report a Tunisian family; their children are affected with DMD. Genetic analysis showed a particular transmission profile indicating a maternal germline mosaicism.

**Patients & Methods:** It is a Tunisian family consisting of two parents, four boys and two girls. Two of the four boys had physical, enzymatic and electrophysiological signs very supportive of DMD. Genetic study allowed an analysis of deletions and duplications of different exons of the dystrophin gene by MLPA (Multiplex Ligation-dependent Probe Amplification). Genotyping by analysis of six microsatellite markers surrounding or inside the dystrophin gene helped to establish the haplotype of each individual and to follow the transmission of haplotype associated with the disease.

**Result:** The MLPA showed in both affected boys, a deletion of the exons 61 and 62 of the Dystrophin gene. However, the indirect study by genotyping revealed the presence of the haplotype stage associated with the disease in three boys with one of which is phenotypically healthy and does not have the deletion responsible for the disease in his two brothers.

**Discussion & Conclusion:** We have a case of germline mosaicism. Indeed, the mother transmitted the same haplotype of the X chromosome to three of her sons of which only 2 are sick and carrying the deletion of exons 61 and 62 of the Dystrophin gene. This is explained by the presence in their germ cells (oocytes), of 2 cell populations; one carries the deletion and the other not, allowing a random transmission of the disease to his sons. This represents a rare case of germline mosaicism confirmed genetically.

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