**HIGHLIGHTS**

* Correlate genotype with phenotype in a family with a novel *GLA* missense mutation, which leading to classical FD;
* The disease caused by p.N34D mutation affected men and women with similar course of the disease;
* p.N34D produces an unstable enzyme that is prematurely degraded in the endoplasmic reticulum, promoting *in vitro* residual activity of 4%;
* Although unstable, the enzyme is catalytically active, responding to treatment with pharmacological chaperone, promoting 37% of residual activity;