A case of multiple angiomas without any angiokeratomas in a female heterozygote with Fabry disease.

Fabry disease is a rare, X-chromosome-linked lysosomal storage disease caused by a deficient alpha-galactosidase A enzyme. The disease manifests primarily in affected hemizygous males and to some extent in heterozygous females ('carrier'). A 45-year-old female Fabry disease patient without angiokeratomas but with numerous angiomas is presented. Her leukocyte alpha-galactosidase A activity was reduced (0.35 nmol/min/mg protein; normal range: 0.4-1). The analysis of her alpha-galactosidase A gene (exon 1-7) showed the transition c.427 G>A. An intrafamilial follow-up search detected a reduced leukocyte alpha-galactosidase A activity in her father, who suffered exclusively from coronary heart disease. Our case report underlines the possible wide range of clinical signs in Fabry disease patients, sometimes complicated by missing typical lesions (e.g. angiokeratomas). In oligosymptomatic Fabry disease cases, genetic analysis is recommended.
Sprache: eng
Print-ISSN: 0004-8380
TUM Einrichtung: r Dermatologie und Allergologie
Occurences:

- Einrichtungen > Fakultäten > Fakultät für Medizin > Kliniken und Institute > Klinik und Poliklinik für Dermatologie und Allergologie > 2010

entries: