



Pigmentary Disorder, Reticulate, With Systemic Manifestations

Alternative Names

PDR
Amyloidosis, Familial Cutaneous

WHO International Classification of Diseases

Diseases of the skin and subcutaneous tissue

OMIM Number

301220

Mode of Inheritance

X-linked

Gene Map Locus

Xp22-p21

Description

The X-linked reticulate pigmentary disorder with systemic manifestations is a rare skin disorder characterized by a hyperpigmentation disorder of the skin. This disorder is mild in females showing brown pigmentation of the skin which follows the lines of Blaschko, while it is severe appearing as reticulate sheets in males. Systemic manifestations are often seen in males. Males may suffer severe gastrointestinal disorders in infancy with failure to thrive and early death. Other manifestations may appear such as corneal dystrophy with severe photophobia or chronic respiratory disease.

Molecular Genetics

The reticulate pigmentary disorder with systemic manifestation is an X-linked disease which has been mapped to a locus in Xp22-p21.

Epidemiology in the Arab World

Lebanon

Megarbane et al. (2005) reported a boy of first cousin parents with X-linked reticulate

pigmentary disorder with systemic manifestations. He showed difficulties in opening his eyes in a lit room, recurrent episodes of bronchitis, a hyperpigmentation of the skin followed by a guttate hypopigmentation, severe photophobia, chronic tearing, a mottled reticulate pattern all over the body, a very high number of melanosomes in the keratinocytes, and many melanophages in the superficial dermis. Molecular analysis of the androgen receptor gene of the mother showed a skewed X inactivation pattern in blood lymphocytes, superior to 95%, which demonstrated her carrier status.

United Arab Emirates

Yahya et al. (1998) conducted a retrospective histopathologic analysis of 490 native kidney biopsies performed on adult patients presenting to four hospitals in the Emirate of Abu Dhabi from 1978 to June 1996. Primary glomerular disease accounted for 77.1% of all biopsies. Of the patients with secondary kidney diseases, 33 (40.7%) had systemic lupus erythematosus, 27 (33.3%) amyloidosis, 14 interstitial nephropathy, and seven diabetic nephropathy.

References

- Megarbane H, Boehm N, Chouery E, Bernard R, Salem N, Halaby E, Levy N, Megarbane A. X-linked reticulate pigmentary layer. Report of a new patient and demonstration of a skewed X-inactivation. *Genet Couns.* 2005; 16(1):85-9.
- Yahya TM, Pingle A, Boobes Y, Pingle S. Analysis of 490 kidney biopsies: data from the United Arab Emirates Renal Diseases Registry. *J Nephrol.* 1998; 11(3):148-50.

Contributors

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