

HUMAN INVASIVE DERMATOPHYTIC DISEASE IS CAUSED BY INBORN ERRORS OF *CARD9*

Auteur: Lanternier, F.; Pathan, S.; Vincent, Q.; Liu, L.; Cypowij, S.; Prando, C.; Migaud, M.; Taibi, L.; Ammar-Khodja, A.; Stambouli, O. Boudghene; Guellil, B.; Jacobs, F.; Goffard, J-C.; Shepers, K.; del Marmol, V.; Bachelez, H.; Michel, L.; Lefranc, G.; Fraitag, S.; Bougnoux, M-E.; Boudia, M.; Lortholary, O.; Abel, L.; Casanova, J-L.; Picard, C.; Grimbacher, B.; Puel, A.

Abstract/Résumé Dermatophytic disease is an invasive, sometimes life-threatening, fungal infection caused by dermatophytes, in which there is extensive cutaneous and subcutaneous tissue involvement, frequent dissemination to the lymph nodes and occasionally to the central nervous system. This condition, which is different from banal superficial dermatophyte infection (dermatophytosis), has mostly been reported in North African consanguineous multiplex families, strongly suggesting a Mendelian genetic etiology. We investigated 13 patients with invasive dermatophytic disease from six unrelated Algerian and Moroccan families. Morbidity rates were high and four of the 13 patients died. No other severe infections were reported in the surviving patients, who were aged 40 to 75 years. We sequenced *CARD9* in the patients. The Algerian patients from five unrelated families had a homozygous Q289X *CARD9* allele, probably due to a founder effect. The Moroccan patients were homozygous for the R101C *CARD9* allele. Both these alleles are rare variants. The familial segregation of these alleles was consistent with autosomal recessive inheritance and complete clinical penetrance. Invasive dermatophytic disease may thus be caused by autosomal recessive *CARD9* deficiency. Inborn errors of immunity should therefore be considered in otherwise healthy patients with unexplained severe fungal disease, including dermatophytic disease in particular.

Keywords/Mots clés : dermatophytic disease is an invasive,

Journal title / Revue : JOURNAL OF CLINICAL IMMUNOLOGY

, 0956-7135, "DOI"

, ?, "issue" ,1 , "volume" , 32

0 , "pp" 94-94, sept-12

WOS:000308728900180

Source: