

MEETING ABSTRACT

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A new CARD9 mutation (R101S) in a Brazilian patient with DEEP dermatophytosis

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Background

Deep dermatophytosis had been described in HIV and immunosuppressed patients. Recently, the association with autosomal recessive CARD9 deficiency was found in individuals previously classified as “immunocompetent”. We describe a new CARD9 mutation associated with dermatophytosis.

Methods

We report a 24-year-old Brazilian male with deep dermatophytosis with *Trichophyton mentagrophytes* isolated from the skin lesions. Opsonophagocytosis of *Candida* was performed. CARD9 was amplified with specific primers.

Results

The symptoms initiated with oral candidiasis at 3 years old, generalized afterwards and treated with oral and local therapy. At 11 years old well delimited, descamative and pruriginous skin lesions appeared; ketoconazole and itraconazole were maintained for 5 years. At 14 years old, the lesions were ulcerative, secretive and painful in the shoulders (15cm of diameter); terbinafine and posaconazole were used without result. His brother presents superficial dermatophytosis. A homozygous mutation in CARD9 exon 3 (R101S) was identified in the patient. His parents, one brother (with superficial dermatophytosis) and one sister are heterozygous for this mutation. Laboratory evaluations showed eosinophilia and high IgE levels; *Candida* killing was clearly impaired in the patient.

Conclusions

This is the first report of CARD9 deficiency in a Brazilian family and the first report of a CARD9 R101S

mutation. A different mutation affecting the same amino-acid (R101C) had been previously described in two Moroccan siblings with deep dermatophytosis.

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