GENETICS AND GENODERMATOSES

EXPANDING MUTATION LANDSCAPE OF BATHING SUIT ICHTHYOSIS IN TUNISIA

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Introduction: Bathing suit ichthyosis (BSI) is a rare variant of autosomal recessive congenital ichthyosis (ARCI), characterized by scales limited to the trunk, sparing the face and limbs. It is mainly due to mutations of the transglutaminase 1 (TGM1) gene.

Objective: The aim of our study was to expand the genotypic spectrum of BSI in Tunisia by identifying recurrent and novel TGM1 mutations.

Materials and methods: A retrospective study was conducted from 2008 to 2017 (10 years). A total of 7 patients with BSI were enrolled in the study from the records of 3 academic medical centers in our country. Each patient underwent targeted sequencing of TGM1 and a detailed clinical history was obtained from their records. Our study was approved by the national ethics committee.

Results: Of the 7 patients, 6 were female and one was male (sex ratio M/F: 1/6). The mean age was 2.14 years (3-48 months). We found 1 novel TGM1 mutation (I304F) in 2 cases and 2 TGM1 mutations (pR315C and pW263X) that have been previously reported in the literature in the remaining patients. All our cases were born with a collodion membrane encasing their entire body. Six patients were born to consanguineous parents (85.7%) and 3 patients (42.8%) originated from the same region. Five cases (71.42%) originated from coastal regions of the country with mild temperate climate while only 2 patients were from the south region of Tunisia with high temperature climate. All patients developed a classic BSI phenotype. There was no significant difference in the severity of this disease between our patients with different genotypes.

Conclusions: Until recently, there has been no genotype-phenotype correlation in BSI. The same mutation of TGM1 could result in either generalized ARCI or BSI. We identified a novel TGM1 mutation (I304F) that was not previously reported in BSI.