Filaggrin Gene Polymorphism Pro478Ser, but Not Loss-of-Function Mutations Mp.Arg501Ter or C.2282del4, Relates with Atopic Dermatitis Severity and Increased Staphylococcal aureus Colonization in Adult Patients

Liliana Rocha1, Cristina Lopes, MD2,3, Susana Fernandes4, Oksana Sokhatska5, Jose Soares6, Freni Tavaria7, Manuela Pintado7, Andre M. Moreira, MD3,9, Luis Delgado, MD8,10, 1Genetics Department, Faculty of Medicine, University of Porto, Portugal, Porto, Portugal, 2Laboratory of Immunology, Basic and Clinical Immunology Unit, Faculty of Medicine, University of Porto, Portugal, Porto, Portugal, 3Allergy Unit, Pedro Hispano Hospital, Matosinhos, Portugal, 4Genetics Department Medical faculty Porto University, 1Immunology Laboratory, Medical Faculty, Porto University, 6Biology and Fine Chemistry Unit, Biotechnology Faculty, Catholic University, Porto, Portugal, 7Biology and Fine Chemistry Unit, Biotechnology Faculty, Catholic University, Portugal, 9Laboratory of Immunology, Basic and Clinical Immunology Unit, Faculty of Medicine, University of Porto, Porto, Portugal, 5Allergy and Clinical Immunology Department, Centro Hospitalar São João, EPE, Porto, Portugal, 10Immunology Lab, Department of Clinical Pathology, Porto, Portugal.


METHODS: In this cross sectional study, data from 73 patients, (30±13 years, 61% female, 77% atopic) with AD for 16±20 years was analyzed. Mutations were analyzed by PCR amplification of exon 3 of FLG gene and Sanger sequencing, disease severity trough SCORAD, allergy by serum levels of IgE, Phadiatop, eosinophil cationic protein and specific IgE to Staphylococcus aureus (SA) enterotoxin A, B, C, TSST and Malassezia spp. Number of colony forming units of staphylococci and SA species in 25 cm2 of poplitea, brachial ceases, interscapular regions were determined. Non-parametric statistic analyses and chi-square were used.

RESULTS: FLG mutations p.Arg501Ter (n=9) and c.2282del4 (n=2) were identified in 14.8% patients and were not associated with AD severity, allergic or microbiological parameters; p.Pro478Ser polymorphism was present in 38% (n=28) of participants and was associated with more severe disease (p=.005), higher colonization with SA (brachial right cease p=.97, popliteal right cease, p=.04, left popliteal cease, p=.02) and had relation with allergy in the subgroup of atopic patients.

CONCLUSIONS: Our study further emphasizes the role of the FLG gene polymorphism p.Pro478Ser, on AD pathogenesis and the IgE response in allergic adult subjects.