

Netherton ´s syndrome defined as a primary immunodeficiency

María Claudia ORTEGA-LÓPEZ

University Hospital of the Santa Fe de Bogotá Foundation, Colombia



Abstract

Son, from parents non consanguineous. Date of birth 11/03/2010 in Panama City. Weight at birth 2900 gm. No known reactions to vaccines. No observable skin anomalies at birth. On the 5th day of life; erythema, peeling face. At 2 months: on an exclusive diet of breast milk; erythema, widespread scaling. At age one, presented paronychia, onychomycosis, recurrent skin infections, bloating, diarrhea with mucus, blood, abdominal pain and sepsis. He underwent an endoscopy with severe esophagitis with eosinophils. He was referred to study for inborn errors of immunity. In 2011 his probably diagnosis was food allergy, enteropathic acrodermatitis, Hyper IgE, Ommen, atopic dermatitis, mycosis, chronic DNT, rickets, low levels of vitamin D, ectropion, septic arthritis, and colitis. We studied specific IgE for food allergy, metabolic analysis, endoscopy, colonoscopy, skin and gas- trointestinal tissue samples, immunological screening, bacterial, viral, fungal cultures. Immunological analysis: CD3: 4.525/68%, CD4: 1939/29%, CD8: 2.592/38%, CD4/CD8:0.7, IgG 602 mg/dL, IgA 42mg/dL, IgM 110mg/dL IgE 223 UI, C3-116 mg/dL, C4-30 mg/dL. HIV: negative. Blood culture: Pseudomonas aeruginosa, Enterococcus faecalis, Staphylococcus aureus, Candida haemulonii, Clostridium difficile. Skin culture: Acinetobacter baumannii, Enterococcus faecalis, Pseudomonas aeruginosa. Urine culture: Pseudomonas aeruginosa, E. coli. Post IGIV treatment: IgA 138-155, IgG 2233-1373, IgM 84-106 IgE 1272. Hair analysis: Trichorrhexis invaginata in hair follicles. At this time, we suspected Netherton ´s Syndrome. Genetic study SPINK5: Chromosome 5q32. Heterozygous c.1258A>G p.Lys420Glu c.1480-3C>T.

Biography

María Claudia Ortega-López has completed her medical and first clinical specialization at the age of 26, from School of Medicine at Universidad Javeriana in Bogotá and her clinical specialization in allergy and clinical immunology at the age of 33 from School of Medicine at Universidad de Sao Paulo Brazil. She is the director of Pediatric Allergy and Clinical Immunology in Hospital Infantil de San José and consultant in Hospital Fundación Santa Fe, in Bogotá. She has published more than 20 papers in distinguish journals and has been serving as a founder- director member of Fundación Alejandra Ortega López.

