GENETICS AND GENODERMATOSES

DIAGNOSTIC ODYSSEY – FROM PROTEIN DIATHESIS TO PITYRIASIS RUBRA PILARIS

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Diagnostic Odyssey – from protein diathesis to pityriasis rubra pilaris.

Introduction: Rare disorders are one of the most important issues in contemporary medicine. The major problem is getting diagnosis, which initially is often erroneous, especially when symptoms are not pathognomonic and develop later in life.

The aim of the study is to present a case of 14 yo patient, in whom pityriasis rubra pilaris due to novel de novo mutation in CARD14 gene (encoding caspase recruitment domain family) was finally diagnosed after 10 years from first clinical symptoms outcome.

Patient and Methods: The patient is 14 years old male. The first skin lesions: erythrodema with desquamation appeared when he was around 4 months. Initially he was treated by dermatologists as protein intolerance (diathesis). Healthy unchanged skin remained only in the area of the nose, the nasolabial fold and the lower lip. The intensity of pruritus was 7 on the VAS scale (0-10).
Subsequently, the patient was suspected of Comèl-Netherton syndrome; his hair showed the presence of trichorrhexis invaginata. Lab tests revealed only the increase in total IgE concentration in the range of 12-14, while other results were within normal ranges.
Next, before being admitted to our center, the patient was diagnosed with psoriasis.
The patient was treated with cyclosporine 50 mg twice daily without improvement and with methotrexate 7.5 mg daily for 3 months without visible improvement.
At the age of 14 yo the clinical suspicion of pityriasis rubra pilaris was suggested. Finally, whole exome sequencing was performed and revealed the novel c.394A>T p.(Ile132Phe) missense mutation in CARD14, that haven't been detected in any parent of the child.

Results and Conclusions: Identification of c.394A>T p.(Ile132Phe) in CARD14 finally confirmed the diagnosis of a rare genodermatoses –PRP.
Whole exome sequencing is a valuable tool in diagnostics of not specific dermatological symptoms.