GENETICS AND GENODERMATOSES

MYH9 AND STEATOCYSTOMA MULTIPLEX SUPPURATIVA: A NEWLY IDENTIFIED MUTATION IN 2 PATIENTS AND THEIR FAMILY MEMBERS

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Background: Steatocystoma multiplex suppurativa (SMS) is a rare skin condition characterized by the presence of multiple subcutaneous cysts and inflamed hidradenitis suppurativa (HS) like lesions. Heterozygous mutations in the KRT17 (keratin 17) gene were described as the cause of the SMS. We report the first mutation in the MYH9 gene identified in 2 patients with SMS.

Observation: Two male patients (patient 1:45 years; patient 2: 37 years) with SMS introduced themselves in our outpatient HS consultation. Both presented with more than 100 non-inflammatory cysts spread over the whole body and several inflammatory foci, such as acute nodules or abscesses in the area of body folds. Patient 1 also suffered from psoriasis and reported severe hearing loss and the existence of lipomas in his mother and brother. Patient 2 reports similar cysts in his father.
Both patients underwent genetic testing, identifying a heterozygous mutation in the MYH9 gene (variant c.5695G>A; p. Glu 1899Lys) following an autosomal dominant inheritance. The mutation was also detected in the mother and brother of patient 1. Consecutive tests confirmed a pathological audiogram and macrothrombocytopenia in patient 2.

Key message: We describe for the first time a mutation in the MYH9 gene in patients with SMS, which encodes a non-muscular myosin involved in cell motility and had previously been linked to a multisystem disorder affecting ears, eyes, thrombocytes, kidneys and liver with high intrafamilial variability. Further studies are needed to investigate the significance of this mutation in patients with SMS and perhaps cystic form of HS. Careful consideration should be given to possible comorbidities in patients with MYH9-associated SMS, such as hearing loss,thrombocytopenia, nephropathy elevated liver enzymes, as well as the possibility of transmission to their offspring in the care and counseling of our patients.