GENETICS AND GENODERMATOSES

NOONAN SYNDROME (LEOPARD SYNDROME) - MULTIPLE LENTIGINES IN DISGUISE

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Background: Noonan syndrome with multiple lentigines (NSML) is a rare condition with approximately 200 reported patients worldwide. The condition, formerly known as Leopard syndrome, affects different organ systems. Diagnosis is based on clinical criteria or on genetic testing.

Observation: We present a 45-year old female patient with multiple lentigines and congenital deafness. NSML had not previously been diagnosed. The patient was 1.53 m tall, had widely spaced eyes (ocular hypertelorism) and low set ears. Although 70-85 % of the patients suffer from sometimes life limiting heart defects, our patient showed no evidence of cardiac involvement (normal echocardiography and electrocardiography). The gynecological status showed no pathological findings. The patient's mother also suffered from multiple lentigines and deafness. According to our knowledge, Melanoma has only been described in three patients and does not seem to be associated disproportionately high with NSML. Because of thousands of small brown macules however, these patients require regular thorough full-body skin examinations. We excised the three largest (up to 5 cm in diameter) and most suspicious brown macules of our patient. Histopathological examination demonstrated junctional nevi.

Key message: Multiple lentigines can be associated with several syndromes such as NSML, Peutz-Jeghers syndrome, Cronkhite-Canada syndrome and Carney Complex. Therefore these individuals demand a careful clinical workup. Noonan syndrome with multiple lentigines is associated with heart defects in 70-85 %. About 80 % of these patients suffer from hypertrophic cardiomyopathy and in up to 20 % pulmonary stenosis has been described. Therefore the individuals need to have regular cardiological check-ups.