GENETICS AND GENODERMATOSES

CORRELATIONS BETWEEN PSORIASIS AND NOONAN SYNDROME

Andreia Mateus Moreira [(1)] *- Eliana Daiha* [(2)]

Universidade Federal Do Esatdo Do Rio De Janeiro, Dermatology, Rio De Janeiro, Brazil [(1)] *- Universidade Federalo Do Estado Do Rio De Janeiro, Dermatology, Rio De Janeiro, Brazil* [(2)]

Background: Pustular psoriasis (PP) is a rare form of psoriasis characterized by an eruption of sterile pustules. Noonan syndrome (NS) is an autosomal dominant disorder and it is one of the most common causes of congenital heart disease. The clinical features vary with age, but typical signs of NS include characteristic facial features with hypertelorism, down-slanting palpebral fissures, chest and spinal deformities, short stature, specific heart defects, learning disabilities and mild mental retardation.

Observation: We describe the case of a woman with features of NS, presented in the last 8 years an eruption of erythematous, annular, and polycyclic lesions, eruptions of small sterile pustules and fine desquamation. Multiple solitary and confluent plaques were present on the torso, axillary and inguinal regions with the clinical appearance of PP and confirmed by biopsy. The patient was treated with antihistamines, topical corticosteroids, and moisturizing lotions, with frequent relapses. After being introduced acytretin 20mg daily, the pacient had good response after 3 months.

Key message: The Ras/MAPK pathway is critical for cell growth, differentiation, senescence, and death. Mutations in genes directly involved in cell proliferation kinase cascades (SOS1, BRAF, KRAS and RAF1) can be associated to a higher frequency of proliferative skin lesions such as hyperkeratosis. Several components of this pathway are related to the promotion of epidermal proliferation and may interfere in the epidermal cell state in skin diseases like psoriasis. NS and other related syndromes may be related to PP because of the common pattern of germ line mutations in the components of the Ras-MAPK pathway. We propose that PP is possibly induced by RAS/MAPK signaling activation in NS; this association can open new lines of research.