Identity Dilemma: Naegeli-Franceschetti-Jadassohn Syndrome

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Introduction

There is too much emphasis on personal identity and huge data are created based upon biometric identification like finger printing, iris pattern identification and DNA typing. Millions of people are subjected to personal identification testing for machine readable document and immigration procedures. As a result of this, certain rare conditions are reported. One of them is Naegeli-Franceschetti-Jadassohn syndrome first described by Oskar Naegeli in 1927 as familiärer Chromatophoren-Naeus in a Swiss family. In 1954, Franceschetti and Jadassohn further analyzed the syndrome as did Itln and colleague in 1993. This case report is first of its kind in our region. 2,3

Case report

A 18 year old young male presented in January 2012 with the unusual complaint of inability to get his computerized national identity card from NADRA authorities. On examination he was found to be having brownish black reticular marking over his neck (Figs 1&2). He was having marfanoid features with smooth skin of both the palms(Fig 3). Thumb impressions of both the hands were taken and were found to have no impressions (Fig 4). Dentition was abnormal (Figure 5)

Fig 1: Brownish black reticular marking on neck

Fig 2: Brownish black reticular marking on neck

Fig 3: Smooth skin of hands

Fig 4: The thumb impressions


Fig 5: Abnormal dentition

**Discussion**

This particular case was diagnosed as Naegeli-Franceschetti-Jadassohn syndrome. The incidence is approximately 1 case in 2-4 million population. It has been reported from Switzerland, Japan, Italy and England. This syndrome is a rare autosomal dominant form of ectodermal dysplasia characterized by reticular skin pigmentation, diminished function of sweat gland, a defective tooth enamel with rough surface and yellow spots, brittle finger nails, hyperkeratosis of palm and sole and absence of finger print lines on the fingers. 3-7

The clinical diagnosis may be confirmed by genetic testing. KRT14 gene is located on chromosome 17q12-21. 8 Studies suggest that NFJ syndrome is caused by frame shift or nonsense mutation in KRT14, leading to early termination of translation or nonsense-mediated degradation of mRNA. Type I keratins have been shown to protect keratinocytes by blocking tumor necrosis factor-alpha proapoptotic signals. Decreased KRT14 has been shown to lead to increased TNF-alpha-induced apoptosis of keratinocytes.9,10 No treatment is effective for this. The lack of dermatoglyphics has been the most consistent clinical feature of this syndrome. Dermatoglyphics comprise variable pattern of epidermal ridges on palmoplantar skin. These ridges form during fetal development in the first trimester. Epidermal ridges first appear at 10 weeks as localized cell proliferation in basal layer of epidermis which form shallow primary ridges that project into the superficial dermis. 11,12

**References**