Phylloid Hypermelanosis: An Unusual Form of Pigmentary Mosaicism

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In this issue, Oiso et al. [1] present an unusual case of pigmentary mosaicism. A 29-year-old man with mental deficiency had hypermelanotic macules arranged in a phylloid pattern. Cytogenetic analysis of peripheral blood lymphocytes showed 3 different aberrant cell types containing either a monocentric or a dicentric ring chromosome 13, or monosomy 13. Remarkably, no normal karyotype could be found in the 30 cells examined. Although the authors did not analyze fibroblasts derived from skin lesions, it is obvious that the phylloid hypermelanosis of this patient reflects cutaneous mosaicism.

The cytogenetic findings documented by Oiso et al. [1] are particularly interesting because aberrations involving chromosome 13 have so far not been described in phylloid hypermelanosis, but only in its counterpart, phylloid hypomelanosis, a disorder that can today be taken as a well-established entity reflecting mosaic trisomy 13 [2–4]. In fact, most cases of phylloid hypomelanosis are caused by mosaic trisomy 13q [2, 5, 6].

Oiso et al. [1] discuss a previous report on phylloid hypermelanosis [7]. Here, I should like to review 2 additional case reports and, at the same time, explain how the story of the phylloid pattern began.

Fig. 1. A case of phylloid hypermelanosis ‘avant la lettre’, as described by Dockx et al. [8] in 1956 (reproduced with permission).

Fig. 2. Phylloid hypermelanosis in a 12-year-old girl with congenital dysplasia of the large vessels [9].

Around 1980, when working at the Department of Dermatology in Münster, I received a short note from our geneticist, Professor Widukind Lenz (1919–1995), the man who had unraveled thalidomide embryopathy and had written a fascinating book on medical genetics that made me enthusiastic about this field of knowledge. He sent me an article, published by Dockx et al. in 1956 [8], which contained a photograph of an unusual pigmentary disorder (fig. 1), and wrote: ‘Dear Mr. Happle. These are *not* the lines of Blaschko, or am I mistaken? Yours sincerely, W. Lenz.’ Indeed, the pigmentary pattern looked bizarre and I did not know how to categorize it. For many years, this article remained in my files but fortunately also in my mind. In 1993, when I proposed the phylloid pattern as a new form of pigmentary mosaicism [9], I wrote: ‘An impressive example of phylloid pattern was documented in 1956 by Dockx et al. in a report on a “congenital pigmentary syndrome close to incontinentia pigmenti (Bloch-Sulzberger) with oligophrenia and multiple osteo-articular malformations”. According to present knowledge this case should not be categorized as incontinentia pigmenti. Most likely it reflects genomic mosaicism.’

In the same article, I described another case of phylloid hypermelanosis in a 12-year-old girl who had an arteria subclavia originating from the aorta descendens (fig. 2) [9]. Cytogenetic analysis of peripheral blood lymphocytes showed a normal karyotype 46,XX.

The number of reports on phylloid hypermelanosis is so far limited, and there are still many questions remaining. Will most cases turn out to be caused by a consistent cytogenetic aberration – as found in phylloid hypomelanosis – or, conversely, is phylloid hypermelanosis a heterogeneous phenotype, i.e. a cutaneous marker of several different types of genomic mosaicism, as found in pigmentary mosaicism of the Ito type? And if so, can we perhaps distinguish different clinical shapes or patterns of phylloid hypermelanosis? For example, some patients show relatively small leaf-like macules [9], whereas other cases are characterized by rather large patches [7, 8]. Or are the unusual melanocytic nevi as noted by Oiso et al. [1] perhaps related to a particular type of phylloid hypermelanosis? It is noteworthy that the patient of Hwang et al. [7] had an unusual hyperpigmented patch showing accentuated hair follicles filled with thick stubby hair, clinically reminiscent of trichostasis spinulosa. This feature has so far not been noted in other cases.

The intriguing report of Oiso et al. [1] will hopefully stimulate further clinical, cytogenetic and molecular research to gain more insight into phylloid hypermelanosis.

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