What's new in pediatric dermatology?

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Pediatric dermatology is a wide and ever changing field. Papers from the last months have been selected focusing on two major topics: infantile hemangiomas and advances in genetic mosaicism. Propranolol has become first line therapy in threatening hemangiomas. A dramatic response with good tolerance has been reported in diffuse hepatic hemangiomas. In a series of eight cases, propranolol treatment resulted not only in improvement of hepatic tumors but also resolution of heart failure and associated hypothyroidism [1]. Ulcerated hemangiomas with prolonged evolution are a common problem, especially on the buttocks. In a non controlled series of infants with ulcerated hemangiomas on propranolol, a rapid response on pain resolution (less than 15 days) and complete healing (less than 1 month) has been found [2]. Early white discoloration as a likely consequence of local ischemia may be an initial sign of ulceration [3], in contrast with late white discoloration in the involuting phase. Ulceration is common in plaque type hemangiomas with minimal growth, which represent a peculiar clinical subtype [4], much more common on the lower body. As previously recognized, it may be associated with anorectal or urogenital malformation, or dysraphism. Besides the acronyms SACRAL and PELVIS, another acronym has been proposes, LUMBAR, which emphasizes on the underlying arterial anomalies [5]. Mosaicism as a consequence of an early post-zygotic mutation can be diagnosed on the skin as segmental or Blaschko-linear diseases. A study of nucleotide and copy number DNA polymorphisms (SNPs and CNVs) in monozygotic twins discordant for schizophrenia has found a high proportion of de novo CNVs (10%), which were absent in parents and had occurred during early developmental mitoses [6]. These events may thus be more common than previously thought. De novo CNVs occur early prior to gastrulation, then remain stable during lifetime in different tissues, resulting in somatic mosaicism [7]. Mosaicism can also occur later during post-embryonic life into adulthood. In a number of hereditary diseases affecting blood cells (primary immunodeficiencies) or the skin (epidermolysis bullosa) partial localized regression due to revertant mosaicism has been found [8]. Revertant skin patches, sometimes with pigmentation, can be seen in patients with junctional or dystrophic epidermolysis bullosa. At the DNA level, mutation reversion is secondary to frameshift restoration or mitotic recombination resulting in a homozygous normal allele. This latter mechanism has recently been evidenced in ichthyosis with confetti, where numerous macules of normal skin on an erythrodermic ichthyotic background result from spontaneous correction of heterozygous keratin 10 gene mutations [9]. The reason why this occurs at a high rate in some patients and not in others who carry mutations in the same genes is not yet known. A strong positive selection of the revertant clones is likely. Revertant mosaicism can be considered as a form of natural gene therapy and might be used for further therapeutic approaches [10].

References


