P7017

Histopathologic findings of microvascular occlusion syndromes

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A wide spectrum of systemic disorders may cause occlusion of the blood vessels. Cutaneous infarction only occurs when numerous vessels in the lower dermis and subcutis are occluded. The differential diagnosis for microvascular syndromes involving the skin is extensive, and the evaluation may be difficult as they can share similar signs and symptoms. Clinical manifestations may include non inflamatory purpura without erythtema, necrotic eschars, branching, cutaneous infarction, retiform purpura or livedo reticularis. Based on pathophysiology, microvascular occlusion syndromes may classified in platelet plugging (eg, heparin necrosis or myeloproliferative disorders), emboli or crystal deposition (eg, cholesterol emboli), vascular coagulopathies, abnormal proteins deposition (eg, cryoproteinemia) or septic embolisms. We present a case series of different microvascular syndromes, including cryoglobulinemia, cholesterol embolism, septic embolism and calciphylaxis. Although these patients showed different clinical presentations among the typical symptoms of microvascular syndromes, the cutaneous biopsy could help in the identification of the underlying process of each case. We would like to hightlight the importance of the histopathological examination in microvascular occlusion syndromes in order to achive an accurate diagnosis.

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P6360

Identification of a novel ECM1 mutation in 2 siblings with lipoid proteinosis

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Lipoid proteinosis is a rare autosomal recessive genodermatosis characterized by the deposition of hyaline material in the skin, mucosa, internal organs, and central nervous system. It is caused by loss-of-function mutations in the extracellular matrix protein 1 gene (ECM1). No effective therapy has been described for this condition yet. A 16-year-old boy was referred to our center with a history of severe long-standing acne-like scarring. The lesions had started in late infancy and had progressed during childhood. The patient's parents were first cousins and a younger brother suffered from a similar problem. The physical examination of both siblings revealed pock-like scars on the face, trunk, and upper extremities as well as beaded papules lining the eyelids. Generalized thickening of the skin was seen in the elder sibling only. Both patients' voices were remarkably hoarse. Investigations including complete blood count, serum biochemistry, EEG, brain MRI, and laryngoscopic examination were all within normal limits. A skin biopsy revealed hyaline deposition in the papillary dermis and surrounding the blood vessels, adnexal epithelia and dermoepidermal junction. Electron microscopy showed thickened vessel walls with embedded pericytes and irregular concentric duplication of the basement membranes. The diagnosis of lipoid proteinosis was confirmed by genetic analysis which revealed a novel homozygous non-sense mutation in exon 9 of the ECM1 gene. Both patients were started on acitretin at 0.5 mg/kg/day. The treatment was well tolerated, but no objective or subjective improvement of the skin lesions or hoarseness were noted after 6 months. The treatment was discontinued. Although previous studies have reported improvement of skin lesions and hoarseness with acitretin, our results were negative. As is the case for all rare diseases, large randomized studies are not feasible and the search for an effective therapeutic option relies on trial and error. Further research into the pathophysiology of lipoid proteinosis will hopefully provide insight to guide future therapeutic trials.

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Human piebaldism

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Human piebaldism is a rare autosomal dominant disorder caused by defective melanoblast proliferation and migration during embryogenesis. Affected individuals present at birth with areas of leukoderma which are achromic and melanocytes are not present in such areas. We present a case of a 4-month-old girl who presented at birth achromic patches on inferior limbs and periumbilical region with small islands of hyperpigmentation and a white forelock. She had no other abnormalities on physical examination. There was no familiar consanguinity. A paternal lineage of affected individuals was observed: great grandfather, grandfather, 3 aunts, 1 uncle and a 3-year old sister showed the same achromic patches. Her father, although affected, did not present achromic areas, but hypochromic one. The familial occurrence, in which more than one generation is affected; with no sex preference, is compatible with an autosomal dominant inheritance with incomplete penetrance and variable expressivity. Molecular studies of this family will help understanding the mechanisms on this rare disorder.

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IQoL: A new ichthyosis-specific measure of quality of life

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Background: Congenital ichtyoses are associated with impaired quality of Life (QoL). In order to explore the facets of this disease and monitor changes in QoL, a specific tool was needed. The aim of this study was therefore to create and validate a specific QoL questionnaire.

Methods: Focus groups worked on a first list of items. After streamlining these items, a prequestionnaire (Q) was drawn up and subjected to a cognitive debriefing. During the validation phase, this Q (together with the DLQI, SF-12, and severity) was sent to patients aged 15 and over, suffering from ichtyosis of varying forms and severities. A shortened version of the Q was designed by removing repetitive items and those identified by the analysis as not having an impact on the score. The validity of the tool was confirmed: both for its structure and one-dimensional nature (Cronbach α), convergent (Spearman correlation) and discriminating validity (Tukey test); α fixed at 5%. The English version of this questionnaire has been validated. Results The initial Q comprised 60 items. During the validation phase, 59 subjects were tested. The shortened version included 32 Items (IQoL-32) and 7 dimensions (Cronbach 0.94). The higher the score, the more impacted is the QoL. IQoL-32 is positively correlated to the DLQI ($P < .0001$) and the SF-12 ($P < .0001$). IQoL-32 is highly correlated to the clinical severity: overall analysis (Spearman ranking: 0.72; $P < .0001$, same for the 7 dimensions (highest correlations: discomfort, pain, interpersonal relations). IQoL-32 demonstrated a higher correlation with VAS compared with the DLQI and the SF-12. It also showed a good discriminating power ($P < .0001$) according to overall severity levels (scores were higher especially when severity levels were too).

Discussion: This study helped design the very first specific questionnaire on congenital ichtyosis. Based on a validated method, the IQoL-32 is simple to use in clinical practice and research. It is more sensitive than DLQI or SF-12 when assessing the QoL of ichtyosis sufferers. Study population included patients with ichtyosis of varying forms and severities. These patients were either receiving hospital outpatient care or were members of the patients' association. This Q can therefore be used in all ichtyosis forms.

Conclusion: This specific QoL scale will be a very useful tool for improving the management of patients with congenital ichtyosis.

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