Hair abnormalities in differential diagnosis of congenital ichthyoses in children

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Congenital ichthyoses are a very heterogeneous group of disorders which include isolated autosomal recessive ichthyosis as well as ichthyosis in different syndromes. Due to the purposes of clinical evaluation of patients, molecular and prenatal diagnostics, doctors tend to narrow this large area of diagnostic search to one congenital disorder and accordingly one gene. We showed a practical meaning of hair abnormalities detection in a differential diagnosis of Netherton syndrome, trichothiodystrophy and autosomal recessive congenital ichthyosis.

Hair on scalp is investigated by means of trichoscopy. Some hairs can have visible abnormalities or be short which forms clinical picture of hypotrichosis. Suspected hairs are depilated for the following light and polarized microscopy.

Sometimes children with Netherton syndrome, trichothiodystrophy and autosomal recessive congenital ichthyosis can have a similar phenotype. Trichorrhexis invaginata and trichoshisis were observed under light microscopy in patient with Netherton syndrome and trichothiodystrophy accordingly. “Tiger tail” pattern of the hair shaft was found under polarized microscopy in the patient with trichothiodystrophy. No hair shaft abnormalities were found in patient with autosomal recessive congenital ichthyosis. Netherton syndrome, trichothiodystrophy type 1 and autosomal recessive congenital ichthyosis type 2 were later confirmed by molecular diagnostic method in our patients. Hair shaft abnormalities can help us to make a clinical diagnosis of congenital ichthyosis as well as other inherited syndromes in children and should be investigated further.