Hair Shaft Abnormalities, Clues to the Diagnosis of Congenital Disorders

Lei Shao, MD, Director of Dermatopathology Service

Hair shaft abnormalities often result from structural changes within the hair fibers and cuticles and they may be associated with congenital syndromes/conditions in children. Hair examination is simple, non-invasive and should be considered as a first line investigation in many such conditions. The technique is uncomplicated and sampling may be carried out in the clinic. Hair samples may be obtained by cutting close to the scalp for the examination of hair shaft abnormalities. Plucked hair is needed if the hair roots are to be examined. The hair can be mounted with mounting medium onto a microscope slide for microscopic examination.

Trichorrhexis invaginata (Bamboo hair) and Netherton’s syndrome

Trichorrhexis invaginata or bamboo hair is a hair shaft abnormality in which the proximal element overlaps the distal element, resulting in a "bamboo-like" or a ball-and-socket joint appearance. Most cases of trichorrhexis invaginata appear to be associated with Netherton’s syndrome.

Netherton's syndrome is an autosomal recessive disease with the combination of ichthyosis, eczema, alopecia and abnormal hair shafts. Erythroderma is usually present within the first 10 days of life, but typical trichorrhexis invaginata might not be present in the first six months. Other reported clinical findings of Netherton’s Syndrome include; mental retardation, seizures, short stature, recurrent infections or IgG abnormalities and aminoaciduria. The condition has now been mapped to 5q32 caused by mutations in the SPINK5 gene, which encodes a serine protease inhibitor. A wide range of mutations are reported including nonsense mutations, frame shift insertions or deletions, and splice site defects with reduced SPINK5 mutant transcript levels.

Pili torti (Kinking hair) and Menkes syndrome

Hair in pili torti is flattened and twisted on its longitudinal axis of the hair shaft. Typically runs of 4-5 twists are found at irregular interval. Pili torti is found in various other syndromes in addition to Menkes syndrome, such as, Bjornstad syndrome and Rapp-Hodgkin ectodermal dysplasia.

Menkes syndrome is an X-linked recessive condition caused by a defect of copper transport and metabolism. The onset of this disease is usually in infancy with lethargy, abnormal temperature regulation, abnormal tone and seizures. The face of an affected individual is characteristic with pallor, full cheeks, tangled eyebrows and a pronounced cupid's bow to the upper lip. The hair may be normal at birth but becomes depigmented, thin and brittle.
Neurological deterioration is progressive and death occurs before the age of 3. An MRI of the brain may show cortical and cerebellar atrophy. The gene maps to Xq13 close to PGK.

**Polarized hair shaft with a tiger tail appearance and trichothiodystrophy**

Trichothiodystrophy (TTD) is a heterogeneous group of autosomal recessive disorders. Patients with TTD have brittle hair and nails, ichthyotic skin, and physical and mental retardation. Approximately half of the patients display photosensitivity, correlated with a nucleotide excision repair defect. However, patients with TTD do not develop the severe skin problems characteristic of xeroderma pigmentosum. The primary clinical phenotype is that of brittle hair and nails owing to a deficiency in a class of sulphur-rich proteins in these tissues. Genetic analysis has implicated that the nucleotide excision repair-deficiency of TTD most often results from mutations in the genes XPB or XPD, encoding helicases of the transcription/repair factor. The light microscopic examination of a TTD patient’s hair reveals trichoschisis with sharply defined transverse fracture across the hair shaft by. Under polarized light, the hair shaft shows alternate light and dark bands giving a “tiger-tail” appearance. It has been noticed recently that such ‘tiger-tail’ patterns are also found in healthy infants. Hence the amino acid analysis quantitating sulfur levels remains a reliable test for TTD.

Other congenital conditions associated with abnormal hair shaft include Chediak-Higashi syndrome which shows abnormal pigment clumping in the hair shaft and Monilethrix characterized by a beaded appearance of the hair due to periodic thinning.

Most of the changes in the hair shaft are not specific and must be interpreted in the light of appropriate clinical information. Examination of scalp hair is simple and noninvasive. It can provide valuable diagnostic information in a range of pediatric congenital conditions.

<table>
<thead>
<tr>
<th>CME Series</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sponsored by Department of Pathology &amp; Laboratory Medicine</td>
</tr>
<tr>
<td><strong>Date:</strong></td>
</tr>
<tr>
<td><strong>Time:</strong></td>
</tr>
<tr>
<td><strong>Location:</strong></td>
</tr>
<tr>
<td><strong>Speaker:</strong></td>
</tr>
<tr>
<td><strong>Topic:</strong></td>
</tr>
</tbody>
</table>