

Alopecia (Congenital) and Hair Shaft Anomalies

Definition / Supporting Information

Children with congenitally sparse, short, or abnormally textured hair may have a range of underlying genetic, endocrine, and metabolic abnormalities. These may be isolated to the hair or part of a syndrome. They may be a manifestation of various types of hair shaft abnormalities. True isolated congenital alopecia is rare and may be inherited as an autosomal dominant trait. Acquired hair loss (alopecia) is described separately.

Essential History

Ask about:

- Hair growth
- Hair loss / breakage
- Abnormal texture of hair
- Family history
- Development of teeth and nails

'Red Flag' Symptoms and Signs

Ask about:

- Sweating / perspiration
 - Children with ectodermal dysplasia may have reduced sweating and can develop fatal hyperthermia.
- Developmental delay
- Photosensitivity
- Faltering growth
- Impairment or loss of hearing or vision
- Recurrent infections

Look for:

- Abnormalities of nails / teeth / eyes
- Skeletal deformities
- Evidence of facial dysmorphism (see Congenital Malformations)
- Faltering growth
- Developmental delay

Differential Diagnosis / Conditions

Ectodermal dysplasia

- Group of related conditions affecting the hair, teeth, and nails
- May be autosomal dominant / recessive or X-linked dominant / recessive
- Symptoms include:
 - Absent / abnormal hair growth
 - Absence / malformation of teeth
 - Inability to perspire
 - Impairment or loss of hearing or vision
 - Recurrent infections
 - Skeletal defects

Other congenital / genetic conditions

- Cartilage–hair hypoplasia
- Orofaciodigital syndrome
- Phenylketonuria
- Homocystinuria
- Congenital hypothyroidism
- Serious chromosomal defects such as trisomy 13
- de Lange's syndrome

Hair shaft abnormalities

- Can occur as an isolated phenomenon or as part of another diagnosis / syndrome
- Monilethrix (beaded hair syndrome)
 - Variable shaft thickness gives a beaded appearance with internodal breakage.
 - Scalp hairs have regularly spaced differences in their circumference, suggesting a chain of beads.
 - Fragile, short, stubble-like growth
 - Associated problems suggestive of a more widespread ectodermal defect include:
 - Cataracts
 - Brittle nails
 - Faulty teeth
- Pili torti (twisted hair)
 - Irregularly spaced twists along the shaft appear flattened.
 - In cross-section, a straight hair appears round, and a curly hair appears oval.

- Both configurations may be seen in a single strand; this can be an important clue to Menke's kinky hair syndrome, a rare association.
- Fragile, short, light 'off-coloured' hair appears spangled as a result of light reflection.
- Texture is coarse and lustreless.
- Presents in young children and may improve in teens
- Menke's kinky hair syndrome
 - X-linked disease
 - Low serum copper level
 - Low serum caeruloplasmin is diagnostic
 - Progressive cerebral degeneration
 - Normal hair at birth replaced by sparse brittle hair like steel wool
- Trichoschisis (clear transverse fracture through the hair shaft)
 - Seen in trichothyodystrophy
 - Recessively inherited DNA repair deficit characterised by brittle, sulfur-deficient hair
 - Hair may also show alternating light and dark bands like a 'tiger's tail' on polarised light microscopy.
 - Sulfur and / or amino acid analysis of the hair is diagnostic
 - Clinically presents with short brittle hair from early infancy
 - May be associated with other features including photosensitivity
- Trichorrhaxis invaginata ('bamboo hair', in which the distal hair herniates into the proximal shaft like a ball and socket; may disarticulate leaving a 'golf tee' deformity)
 - Associated with Netherton's syndrome
 - Recessive deficiency of a steroid sulfatase inhibitor
 - Presents in infancy with short brittle sparse hair, significant failure to thrive, and abnormally dry and fragile skin
 - Susceptibility to infection and atopy develop

Investigations

To be undertaken by specialist practitioners (eg, Paediatric Dermatology Team):

- Light microscopic analysis
- Serum copper
- Thyroid function
 - Congenital hypothyroidism
 - Plasma and urine amino acids and organic acids
 - Phenylketonuria
 - Homocystinuria

Treatment Approach

Treatment depends on the underlying cause.

To be undertaken by specialist practitioners (eg, Emergency Department / Paediatric / Paediatric Dermatology Team(s)):

- Plastic surgery
 - Expertise should be sought for consideration of hair transplants.

When to Refer

Refer all cases of congenital alopecia and hair shaft anomalies to specialist practitioners (eg, Paediatric / Paediatric Dermatology Team(s))

- Refer urgently children with history of reduced sweating

‘Safety Netting’ Advice

Advise families of children with potential ectodermal dysplasia how to prevent and recognise hyperthermia.

Resources

Medical Decision Support

Lewis-Jones S. [Paediatric Dermatology](#). Oxford Specialist Handbooks in Paediatrics. Oxford University Press, 2010.

Suggested Resources

****Please note: these resources include links to external websites. These resources may not have national accreditation and therefore PCO UK cannot guarantee the accuracy of the content.***

[George’s Story](#) (Web page), Ectodermal Dysplasia Society

[Types of alopecia](#) (Web page), Alopecia UK

Acknowledgements

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Paediatric Specialty Group: [British Society for Paediatric Dermatology](#)

Update information

Created: 2017

Date last updated: 2020

Next review due: 2023