Cysts and Disorders of the Hair

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No Conflicts to Disclose
Hair Anatomy
Keratins 6a, 16 are expressed in outer root sheath.
Hair Anatomy

- VL=vitreous layer, AKA “glassy Membrane”
- Outer root sheath (glycogenated)
- Inner root sheath
- Medulla
- Cuticle
- Cortex
Hair Anatomy – Vertical Section

Elston D., Ferringer T. (2014). Dermatopathology, 2nd ed
Hair Anatomy – Vertical Section

Elston D., Ferringer T. (2014). Dermatopathology, 2nd ed
Hair Anatomy – Transverse Section
Hair Anatomy Regions

**Infundibulum:**
- epidermis to the insertion of the sebaceous gland

**Isthmus:**
- opening of the sebaceous glands to attachment of arrector pili muscle
Hair Anatomy Regions


**Infundibulum:**
- Keratinizes with granular layer

**Isthmus:**
- Contains region known as the “bulge”
- Keratinizes with absence of granular layer = trichilemmal keratinization
- Inner root sheath is lost at this level
- Outer root sheath develops a corrugated, dense pink cornified layer
Regions of Importance

- **Bulge** = segment of outer root sheath at the arrector pili insertion; epithelial stem cells reside here
- **Bulb** = melanocytes generate hair color; location of inflammatory infiltrate in alopecia areata
- **Critical line of Auber** = widest diameter of lower portion of hair follicle; below line of Auber is where the bulk of mitotic activity occurs
Anatomy of Hair Growth Cycle

Telogen hair

Elston D., Ferringer T. (2014). Dermatopathology, 2nd ed
Anagen Hair

M=matrix; I=inner root sheath; O=outer root sheath

Elston D., Ferringer T. (2014). Dermatopathology, 2nd ed
Catagen/Telogen Hair

Elston D., Ferringer T. (2014). Dermatopathology, 2nd ed
Vellus Hair

Non-Scarring Alopecias
Androgenetic Alopecia

- Androgen-dependent, resulting from the conversion of scalp terminal hairs into miniaturized vellus hairs in a characteristic pattern.
- Type II 5-alpha reductase enzyme activity and DHT levels are increased in AGA.
- Histology shows normal number of follicles, increased vellus and telogen hairs, no significant inflammation.
- Treat with minoxidil, finasteride, dutasteride, or hair transplantation.
Telogen Effluvium

• Caused by pregnancy, malnutrition and other stressors that trigger a large numbers of hairs to enter the telogen phase simultaneously.
• Most common drug causes: retinoids, anticonvulsants, antithyroid medications, anticoagulants, lithium, interferons, and β-adrenergic blocking agents.
• Hair loss begins about 3 months after a definable precipitating event.
• Hair pull positive for 2 or more telogen hairs.
Trichotillomania

- More common in females and in children.
- Hair is pulled causing patchy or full alopecia; often have bizarre shapes, irregular borders, and contain hairs of varying lengths.
- Histology shows trichomalacia, pigmented hair casts, and empty follicles.
- Treat with behavioral modification.
- Clomipramine first line pharmacologic.
Alopecia Areata

- Presents as round or oval, non-scarring patches of hair loss; other presentations include totalis, universalis, and ophiasis patterns.
- Exclamation point hairs and yellow dots on trichoscopy.
- Most common nail change is pitting.
- Histology shows lymphocytic inflammation in “swarm of bees” pattern around lower portion of follicle.
- Treatment: ILK, minoxidil, topical anthralin, and combination therapy.
Temporal Triangular Alopecia

• Present at birth or acquired usually within the first decade of life.
• Clinically, lancet shaped patch of alopecia, usually in the temporal region(s).
• Very fine vellus hairs can be seen with magnification
Anagen Effluvium

- Sudden onset loss of anagen hairs.
- Triggered by chemotherapy, radiation, or chemicals (thallium, arsenic)
Other Non-Scarring Alopecias

• Post-operative (pressure induced): related to prolonged pressure, usually on the occipital scalp during surgery.
• Psoriasis associated alopecia: occurs with the shedding of thick psoriatic plaques; may also be related to TNF alpha inhibitors.
Scarring Alopecias
Central Centrifugal Cicatricial Alopecia

- Follicular degeneration from premature desquamation of inner root sheath
- Often seen in African-American women, with follicular loss on the crown or vertex of the scalp, expanding in a symmetric, centrifugal fashion
- Pathology: Premature desquamation of IRS, mononuclear infiltrate at isthmus, eccentric epithelial atrophy, concentric lamellar fibroplasia of affected follicles
- Treatment: Doxycycline; potent topical corticosteroid
Central Centrifugal Cicatricial Alopecia

Fig 2. Central, centrifugal cicatricial alopecia. Symmetrical alopecia centered on the crown/vertex of the scalp.

Fig 3. Central, centrifugal cicatricial alopecia. Horizontal (A) and vertical (B) sections showing premature desquamation of the inner root sheath, eccentric epithelial atrophy, concentric lanellar fibroplasia, and chronic perifolicular inflammation. (Hematoxylin–eosin stain.)
Lichen Planopilaris

- Related to lichen planus; etiology remains unknown
  - >50% associated with cutaneous or oral LP
- Scattered foci of hair loss associated with perifollicular erythema, follicular spines and scarring; often pruritus and tenderness
- Pathology: Band-like lichenoid interface dermatitis of superficial follicular epithelium, affecting upper portion of hair follicle
- Treatment: Antimalarials, topical/intralesional/oral corticosteroids
Lichen Planopilaris

- **Frontal Fibrosing Alopecia** is a distinctive clinical variant of LPP affecting frontotemporal hairline and eyebrows.
Discoid Lupus Erythematosus

- Alopecia with erythema, epidermal atrophy, dilated and plugged follicular ostia
- Pathology: Vacuolar interface of epidermis and follicular epithelium, superficial and deep inflammation with dermal mucin
- Treatment: Oral antimalarials, topical/intralesional/oral corticosteroids
- 50% of patients with DLE have scalp involvement
Acne Keloidalis

- Possible foreign body reaction to hair fragments or ingrown hairs
- Follicular papules/pustules that can progress to keloidal papules with surrounding alopecia on occiput scalp/posterior neck
- Pathology: lymphocytic/plasmacytic inflammation at isthmus/lower infundibulum with lamellar fibroplasia and loss of sebaceous glands; total follicular destruction in advanced disease
- Treatment: Topical steroids, oral antibiotics, surgical excision
Dissecting Cellulitis of the Scalp

- Firm nodules on vertex and upper occiput develop into boggy oval and linear ridges with purulent, foul-smelling drainage though minimal associated pain
- Pathology: Perifollicular lymphocytic/neutrophilic inflammation affecting lower half of dermis and subcutis; end stage with loss of follicles
- Treatment: Oral isotretinoin, intralesional corticosteroids, oral antibiotics, TNFα inhibitors, surgery
End-Stage Traction Alopecia

- Prolonged traction
- Commonly seen in African American women with scarring alopecia of bitemporal and frontal scalp
  - Lag period of a decade or more exists between traction and hair loss
- Pathology: Total number of hairs markedly decreased, persistent sebaceous glands, columns of connective tissue replacing former follicles
- Treatment: adopt alternative hair practices, topical minoxidil, ILK, hair transplant
Pseudopelade of Brocq

- Not a distinct disease - pattern of end stage alopecia of other various forms of cicatricial alopecia
- Leads to asymptomatic, irregularly shaped, atrophic patches of alopecia
- Pathology: Atrophy, loss of sebaceous epithelium, fibrosis with absent hair follicles
- No successful treatment
Other Cicatricial Alopecias

- **Aplasia Cutis Congenita**: disruption of intrauterine skin development causing congenital absence of skin/subcutis leading to atrophic, coin-sized alopecia.

- **Keratosis Follicularis Spinulosa Devalcans**: X-linked recessive disorder of abnormal follicular keratinization seen in childhood with alopecia of scalp, eyebrows and eyelashes; remits in puberty.
Biopsy Techniques of the Hair
Hair/Scalp Biopsy - Indications

• A common procedure used to evaluate the histopathologic processes that can present in many clinical ways.

• May be used to assist with the diagnosis of scarring and non-scarring alopecias, effluvium conditions, infectious diseases of the hair and scalp, hair shaft abnormalities, and many others.

• Most often achieved by a “punch” biopsy technique, but in certain conditions the hair shafts can be:
  • Pulled/Plucked – Telogen/Anagen Effluvium
  • Cut – Nits, Hair shaft anomalies (trichorrhexis nodosa, pili torti, trichothiodystrophy, etc.)
Hair/Scalp Biopsy – Lesion Selection

• As a general rule with any biopsy, the area selected for sampling should be from fresh, but well-developed lesional tissue.
  • For inflammatory conditions involving the hair, areas of active inflammation, erythema, or edema should be chosen.
  • For scarring conditions of the hair or scalp, the peripheral edge of the involved tissue is more likely to provide meaningful pathological changes.
  • For non-inflammatory conditions (i.e. androgenetic alopecia), any involved tissue may be sampled.
  • For chronic diseases, older lesions should be selected
Hair/Scalp Biopsy – Lesion Selection

• On the face, there are a few areas that should be recognized as potentially dangerous. These areas correspond to arteries that lie superficially and may be transected during the procedure.
  • Temple – lateral to the eyebrow (temporal artery)
  • Supraorbital notch – medial brow (supraorbital artery)
Hair/Scalp Biopsy Techniques: Skin Preparation and Local Anesthesia

• After site selection, the area must be adequately prepped and anesthetized.
• Local skin preparation can be achieved with alcohol, chlorhexidine gluconate, and iodine formulations.
• Lidocaine is the most commonly used local anesthetic for skin biopsy, usually delivered by local infiltration.
  • For scalp biopsies especially, lidocaine mixed with epinephrine may offer better hemostasis than plain lidocaine.
  • Approximately 10 minutes should be given after local infiltration of lido + epi to maximize the epinephrine’s vasoconstrictive effects.
• EMLA achieves ~5mm depth after 2 hours of occlusion.
Punch Biopsy: Considerations and Technique

In general, 4mm punch sizes and larger allow the tissue to be transected by the technicians according to the pathologists preference.

- Vertical transection
- Horizontal transection
Punch Biopsy: Considerations and Technique

- Care must be taken to ensure that enough tissue is sampled to show representative changes.
  - In conditions affecting the hair, this often requires sampling down to the subcutaneous fat, as many of the inflammatory processes involve the hair bulb.
  - The necessary depth depends on anatomic site, age of the patient, nature of the disease, etc.
  - Occasionally a 6mm punch is required to obtain the subcutaneous fat.
- The punch should be pushed into the skin and rotated in one direction, as back-and-forth twisting may shear the epidermis completely off.
- Once the subcutaneous plane has been reached, the clinician should feel a decrease in resistance in the downward direction.
Punch Biopsy: Considerations and Technique

• Once the punch has been advanced to the appropriate depth, care must be taken to ensure the specimen:
  • 1. contains sufficient deep tissue
  • 2. is not damaged by forceps during removal
• To accomplish these, the following may be employed:
  • 1. use fine, sharp surgical scissors to carefully separate the base of the punch from the underlying tissue (subcutis).³
  • 2. gently, use smooth-tipped forceps to grasp the specimen (teeth on forceps may cause crush artifact).
  • 3. use the lidocaine needle to “spear” the specimen and retract it from the base

“Spearing” technique using needle from lidocaine syringe – minimizing crush artifact.
What is Crush Artifact?

- Aggressive handling of the biopsy specimen during extraction can damage the tissue, rendering the histologic evaluation difficult or impossible to interpret.
Punch Biopsy Technique – Hemostasis and Closure

- After the specimen has been obtained and placed in the correctly labeled container, the defect may require:
  - Hemostasis (electrocautery, aluminum chloride, Monsel’s solution) – care should be taken to minimize the amount of epidermis damaged by electrocautery, which may cause scarring.
  - Closure (usually for punch biopsy sites > 2-3mm)
    - Simple interrupted sutures usually suffice
    - A horizontal mattress suture may be used to stop bleeding.
    - A vertical mattress suture may help to draw edges together in larger (i.e. 6-8mm) defects if there is little tissue mobility.
    - On the scalp, prolene 3-0 to 4-0 is usually sufficient, and the blue color makes the material easier to identify against a background of hair (nylon can be difficult to find)
HAIR SHAFT ABNORMALITIES
Hair Shaft Abnormalities
WITH Increased Fragility

Trichorrhexis Nodosa: Incomplete fracture with frayed ends resembling two paint brushes against each other.

- Seen in Menkes disease (ATP7a mutation), trichothiodystrophy (ERCC2, ERCC3 mutation), Netherton’s syndrome (SPINK5 mutation), arginosuccinic aciduria (arginosuccinate lyase deficiency), Citrullinemia (argininosuccinic acid synthetase deficiency)
- Most common hair shaft defect

Trichothiodystrophy: Sulfur-deficient hair with alternating light and dark bands under polarizing light

Trichoschisis: Clean transverse fracture of hair shaft
- Seen in trichothiodystrophy
Hair Shaft Abnormalities WITH Increased Fragility

- Trichorrhexis Invaginata: Ball & socket or collapsible telescope; “bamboo hair”.
  - Seen in Netherton’s syndrome

- Pili Torti: Hair fibers flattened and twisted at 180 degree angles.
  - Seen in Bjornstad syndrome (BCS1-like protein deficiency), Crandall syndrome, Menkes disease, Netherton’s syndrome
Hair Shaft Abnormalities WITH Increased Fragility

• Monilethrix: Beaded appearance of hair due to periodic thinning of hair shaft.
  • AD/hHB6, hHB1
  • AR/Desmoglein 4
  • Patients normal at birth, but develop short, brittle hair within a few months
Hair Shaft Abnormalities WITHOUT Increased Fragility

• Pili Trianguli et canaliculi: Premature keratinization of a triangular-shaped internal root sheath; “spun-glass hair”.
  • Prominent longitudinal groove along the long axis of the hair
  • Seen in Uncombable Hair Syndrome- hair appears dry, dull, frizzy, short, and light in color; unmanageable
Hair Shaft Abnormalities WITHOUT Increased Fragility

• Pili Annulati: Alternating light and dark bands seen with reflected light
  • Light bands due to abnormal air-filled cavities

• Loose Anagen Hair: Anagen hairs without inner root sheath; ruffled proximal cuticle
  • Seen in loose anagen hair syndrome- hair is easily pulled from the scalp
Hair Shaft Abnormalities WITHOUT Increased Fragility

• Wooly hair: Tightly curled hair; may see axial twisting, breaks, and splitting.
  • Seen in Naxos disease (Plakoglobin mutation), Carvajal syndrome (Desmoplakin mutation), Wooly hair and skin fragility syndrome (Desmoplakin mutation), Diffuse partial wooly hair

• Curly hair: Large loose spiral hair
  • Seen in CHAND syndrome, Trichodentoosseous syndrome (DLX3 mutation), Costello syndrome (HRAS mutation), Noonan syndrome (PTPN11 mutation)
Hair Shaft Abnormalities WITHOUT Increased Fragility

- Trichoptilosis: Longitudinal splits in hair shaft due to trauma
- Trichonodosis: Knots develop in curly hair due to combing/rustling of hair
- Pili bifurcati: Two hairs, which occupy the same follicle, bifurcate and then rejoin
- Pili multigemini: Multiple hair shafts from one papilla
Cysts

- A cyst is a walled-off sub-epidermal cavity filled with fluid, keratin, or mucin.

- Cysts can be classified by anatomic location, embryologic derivation or histologic features.

- 3 categories for classification:
  - Stratified squamous epithelium
  - Non-stratified squamous epithelium
  - Absence of epithelium
Derivation
Cysts with an Epithelial Lining
Epidermal Inclusion Cyst

- Common cyst found mostly on face and upper trunk
- Arises from the follicular infundibulum
- There is a possible genetic predisposition; Gardner's Syndrome, Nevoid Basal Cell Carcinoma Syndrome, Pachyonychia Congenita
- Lined by stratified squamous epithelium with granular layer and contains lamellated keratin
Milia/Milium

- Arise from vellus hairs
- Common in children
- Common on face
- Widespread distribution found in Hereditary Trichodysplasia (Marie-Unna Hypotrichosis); Oral-Facial-Digital Syndrome Type 1; Rombo Syndrome; and Bazex Syndrome
Pilar Cyst

- Clinically similar to EIC, but located on the scalp
- May be inherited as an autosomal dominant trait
- Frequently multiple
Lined by stratified squamous epithelium with no granular layer; cyst contains homogenized keratin
Proliferating Trichilemmal Cyst

- Slow-growing nodule on scalp
- May increase in size
- Well-circumscribed nodule in the deep dermis with cystic and solid patterns; neoplastic cells with mitotic figures; tumor with pushing margins
Vellus Hair Cysts

- Commonly found on the trunk/chest as dome-shaped skin colored to pigmented papules
- May be inherited an AD fashion
- DDx: eruptive vellus hair cysts
- May be seen in the setting of Pachyonychia Congenita type 2
Lined by squamous epithelium with granular layer with contents lamellated keratin and vellus hairs
Steatocystoma

- Dermal skin-colored to yellow nodule that may drain oily fluid punctured
- Common on chest, axilla, and groin
- Can be inherited as an autosomal dominant condition called Steatocystoma Multiplex
- Seen in conjunction with eruptive hair cysts and in Pachyonychia Congenita
- Cyst lined by stratified squamous epithelium with granular layer with thin, corrugated eosinophilic cuticle with adjacent sebaceous lobules
Dermoid Cyst

- Congenital cyst most often found on the face (lateral eyebrow)
- Lined by squamous epithelium with granular layer with cysts contain lamellated keratin; multiple pilosebaceous units near cyst lining
Hidrocystoma

- Skin colored to translucent or even blue cysts on the face
- Can be classified as Eccrine or Apocrine
- Associated with: Ectodermal Dysplasia; Schopf-Schulz-Passarge Syndrome
- Cyst lined by layers of epithelial cells
Bronchogenic Cyst

- Most commonly found in the suprasternal notch at birth
- Formed from trapped respiratory epithelium of the trachea during embryologic development
- Lined by pseudostratified ciliated columnar epithelium with goblet cells; cyst lining often surrounded by smooth muscle, cartilage, mucous glands, or lymphoid follicles
Thyroglossal Duct Cyst

- Represent failed migration/ remnant of the thyroglossal duct
- Commonly found on the midline neck
Lined with cuboidal, columnar, or stratified squamous epithelium containing characteristic thyroid follicles in cyst wall
Branchial Cleft Cyst

- Often present in the second or third decades of life
- Occurs along the SCM, pre-auricular area, or the mandible
- Due to incomplete involution of branchial cleft structures
- Consider CT or MRI before removal
Lined by stratified squamous epithelium or pseudostratified ciliated columnar epithelium surrounding lymphoid tissue
Cutaneous Ciliated Cyst

- Common on the thighs of young women
- Can be found on the buttocks
- Epithelial lining is cuboidal or columnar with cilia on the surface and collagen and vessels deep to the wall
Median Raphe Cyst

- Occur on the ventral penis near the glans in young men
- Thought to be a remnant of the urethral epithelium
Pseudostratified columnar epithelium with or without mucinous cells
Other

- Pigmented follicular
- Follicular Hybrid
- Ciliated Cyst of the Vulva
Cysts without an Epithelial Lining
Digital Myxoid Cyst

- Most commonly found on the dorsal distal phalanx of the fingers
- Drain clear gelatinous material if punctured
- Can have an underlying connection to a joint space
- Often traumatizes the nail matrix causing nail changes
Not a true cyst since it is devoid of lining; consists of a large pool of mucin containing spindle/stellate fibroblasts
Pseudocyst of the Auricle

- Usually presents as a painless swelling of the scaphoid fossa in middle-aged men
Pilonidal Cysts

Note hair shafts and granulation tissue
Mucocele

- Caused by a disruption of the minor salivary gland ducts
- Commonly located on the lower labial mucosa
- Accumulation of mucinous material can illicit inflammation
Ganglion Cyst

- Large mucinous filled cysts attached to a tendon sheath or joint capsule but not communicating with the joint space
- Mucin is thought to be produced from fibroblasts
- Found on volar wrists, dorsal wrist, fingers, feet or knees
Metaplastic Synovial Cyst

- Occurs at sites of trauma or surgery
- May be lined by reactive cuboidal or epithelioid cells
- Villous projections into cystic cavity
- Frequently connected to the epidermis through fistulous tracts
References

- Bolduc, Chantal, MD, Leonard Sperling, MD, and Jerry Shapiro, MD. "Primary Cicatricial Alopecia." Journal of the American Academy of Dermatology 75.6 (2016): 1081-111. Print. (2)
THANK YOU!