Hair Shaft Defects

Hair Shaft Defects

- Clinical presentation is variable
  - Childhood or adulthood
  - Inherited or acquired
  - Patchy or diffuse alopecia
  - Isolated or associated anomalies

Hair Shaft Defects

- Increased fragility
  - Monilethrix
  - Pili torti
  - Menkes' Kinky Hair
  - Netherton's
  - Trichorrhexis Nodosa
  - Trichothiodystrophy
- Normal Strength
  - Pili Annulati
  - Wooly Hair
  - Straight Hair Nevus
  - Progressive Kinking
  - Uncombable Hair
  - Loose Anagen

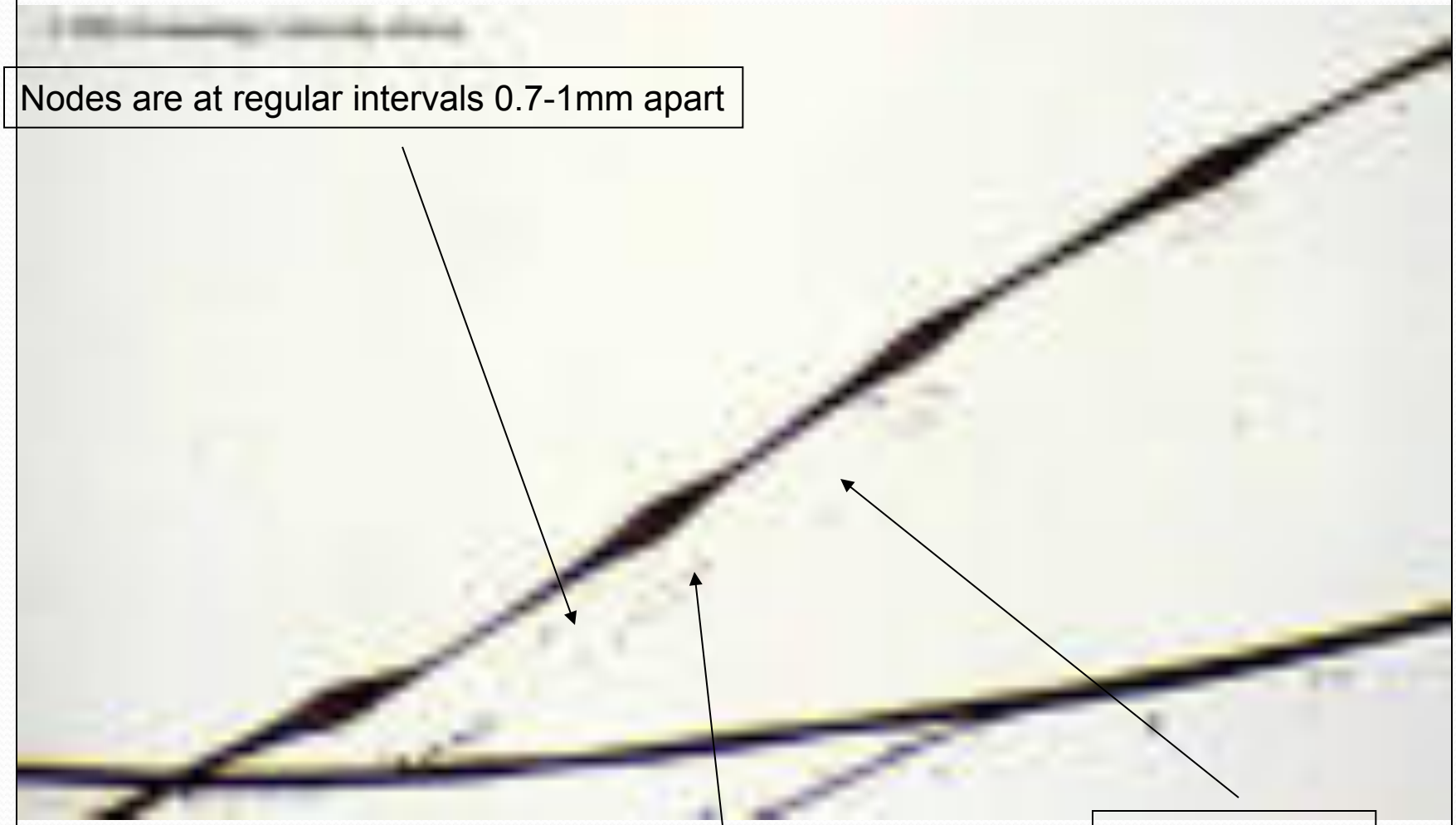
Hair Shaft Defects

- Terminology
  - “tricho”: pertaining to hair
  - “pilo”: pertaining to hair
  - “thio”: prefix denoting sulfur
  - “monil”: necklace
  - Suffix “schisis”: cleavage
  - Suffix “ptilosis”: feather
  - Suffix “clasis”: break off

Increased Fragility

- Monilethrix
  - Genetics
    - Autosomal dominant
    - Mutation in Hhb1 and Hhb6
  - Clinical
    - In childhood w/patchy alopecia and dry, lusterless hair
    - Follicular keratosis at nape of neck; KP common
    - Stable, but may improve at puberty
  - Pathology
    - "beads" every 0.7-1mm apart
    - No medulla at internodes
  - Associated defects: rare case reports (cataracts, oligophrenia)

Monilethrix



Monilethrix

Lusterless, brittle hair. Patchy alopecia due to breakage.

Increased Fragility

- Pili Torti
  - Genetics
    - Autosomal dominant
  - Clinical
    - Childhood; hair won't grow beyond a few inches
  - Pathology
    - "Twisted look": hairs are flattened, and rotated 180 at irregular intervals
  - Associated defects:
    - Menkes', Bjornstad's, ectodermal dysplasia

Pili Torti

Increased Fragility

- Pili Torti (cont)
  - Menkes Kinky Hair
    - Genetics
      - X-linked recessive (males only)
      - Defect in absorption of copper (copper transporting ATPase)
    - Clinical
      - Nl at birth, then replaced by depigmented, steely hair
      - Mental retardation, cerebral degeneration
      - Classic facies, “cupid’s bow” lip, doughy redundant skin
      - Death in early childhood
    - Pathology
      - Pili torti (classic), and trichorrhexis nodosa
    - Associations
      - Female carriers may show pili torti

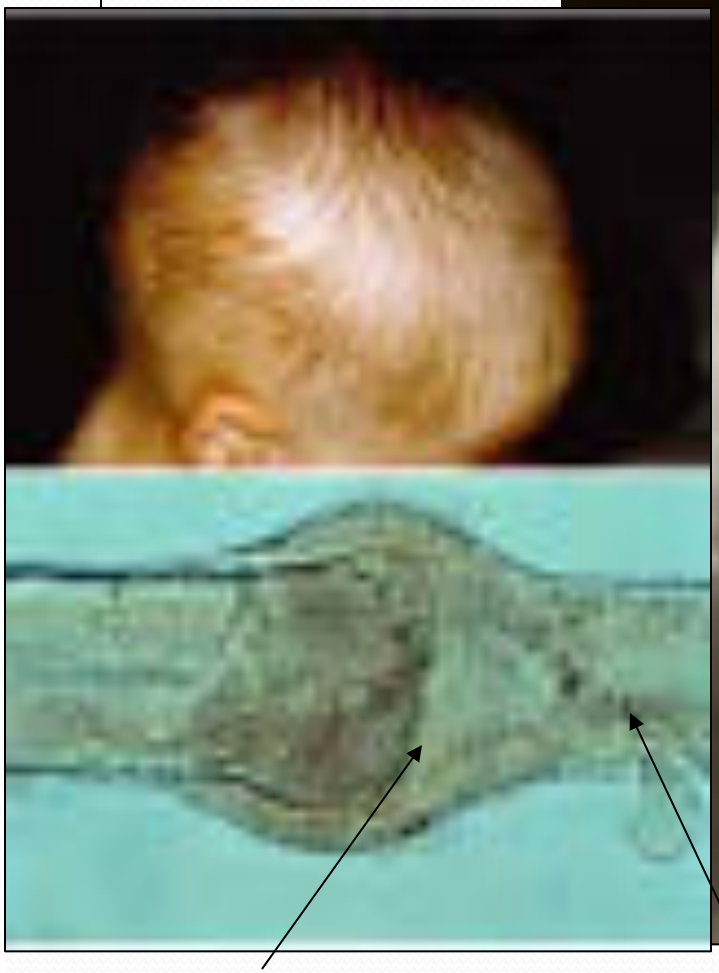
Increased Fragility

- Pili Torti (cont)
  - Bjornstad's
    - Genetics
      - AR and AD reported
    - Clinical
      - Present in early childhood
      - Short brittle hair and bilateral senorineural deafness
    - Pathology
      - Pili torti
    - Associations
      - Crandell syndrome: pili torti, deafness, hypogonadism

Increased Fragility

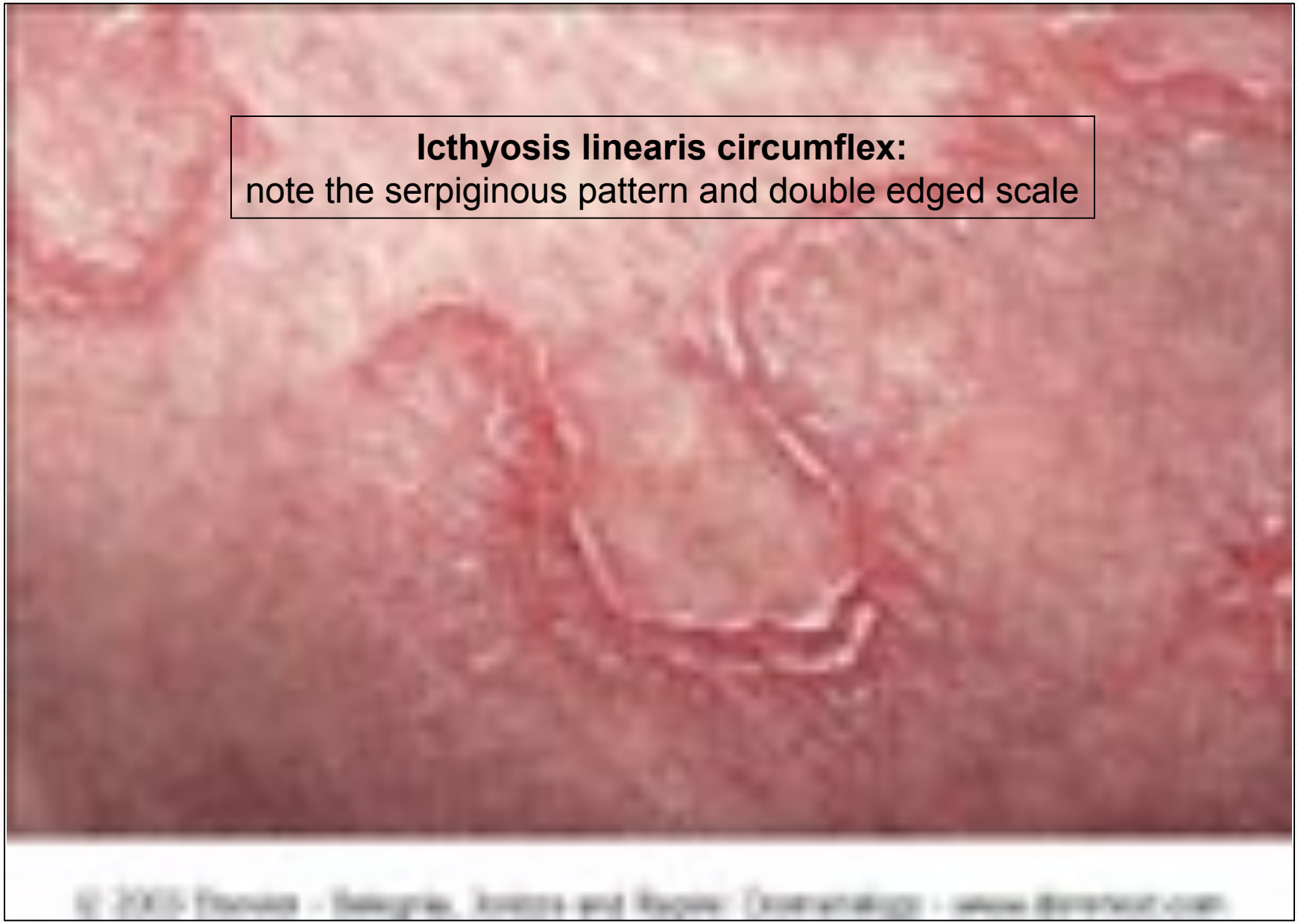
- Netherton's
  - Genetics
    - Autosomal recessive
  - Clinical
    - Born erythrodermic; develop classic icthyosis linearis circumflexa scale
    - Short sparse hair that does not grow; all affected
    - Anaphylactic food reactions
    - Nl intelligence, nl life span
  - Pathology
    - Trichorrhexis invaginata (ball-socket, bamboo)
  - Associations
    - May also have trichorrhexis nodosa and pili torti

Netherton's syndrome: trichorrhexis invaginata



"Ball"

"Socket"



Icthyosis linearis circumflex:
note the serpiginous pattern and double edged scale

Increased Fragility

- Trichorrhexis nodosa
  - Genetics
    - Most common shaft defect, least specific
    - Unclear if genetic predisposition vs. all trauma
  - Clinical:
    - Broken hairs, some alopecia
  - Pathology
    - Small white nodes at irregular intervals along shaft: disrupted cuticle prone to breakage
      - "Paint-brush" or "broomstick" appearance
  - Associations
    - Arginosuccinic aciduria: urea cycle defect
    - Proximal = African Americans; Distal = Caucasian

Trichorrhexis nodosa

Increased Fragility

- Trichothiodystrophy (Tay syndrome)
  - Genetics
    - Autosomal recessive, defect in sulfur proteins
  - Clinical
    - Variable icthyosis
    - Short, brittle hair
      - low sulfur/cystine content (<50% of normal)
  - Pathology
    - Nl/flattened or trischoschisis on light microscopy
    - “tiger-tail” appearance on polarizing microscopy
  - Associations
    - Various other associations compose “trichothiodystrophy syndromes”

Increased Fragility

- Trichothiodystrophy syndromes
  - Unclear if all part of similar entity
  - Low sulfur content is constant
    - IBIDS: icthyosis, brittle hair, intellectual impairment, decreased fertility, short stature
    - PIBIDS: photosensitivity
      - Same DNA repair defect as in XP type D
      - Do NOT develop cancers?!

Trichoschisis: clean break in shaft

“Tiger-tail” on polarizing microscopy

Normal Strength

- Pili Annulati
  - Genetics
    - Autosomal dominant
  - Clinical
    - Short hair with decreased growth rate
    - Shiny appearing, “attractive”
  - Pathology
    - Alternating dark and light band on light microscopy
      - Light bands due to air in the cortex
  - Associations
    - none

Bright, shiny hair that reflects light

Alternating bands on *light* microscopy

Normal Strength

- Wooly Hair
  - Genetics
    - 3 types:
      - autosomal dominant, autosomal recessive: present at birth
      - localized (wooly hair nevus)
  - Clinical
    - Tightly coiled hair in a person NOT of African background
  - Pathology
    - Tightly coiled hair shaft
    - Ovoid in cross section (not round)
  - Associations
    - none

Normal Strength

- Wooly hair
  - Wooly Hair Nevus
    - Genetics
      - No known inheritance
    - Clinical
      - Childhood
      - tightly curled hair in a circumscribed patch on scalp
      - Hair finer, lighter
    - Pathology
      - Wooly Hair
    - Associations
      - Verrucous or epidermal nevus elsewhere


Discrete, course patch amidst fine straight hair

Normal Strength

- Acquired Progressive Kinking of Hair
  - Genetics
    - No genetic pattern
  - Clinical
    - Onset in early adulthood; M>F
    - Gradual curling and darkening of the hairs, like “pubic hair”
  - Pathology
    - Shortened anagen phase
    - Shaft is curled
  - Associations
    - “Whisker Hair”:
      - a variant in some men; the kinky hairs are located around the scalp margins
      - Subsequently develop androgenetic alopecia

Normal Strength

- Straight Hair Nevus
  - Genetics
    - No known defect or inheritance
  - Clinical
    - Circumscribed area of straight hair in a black patient
  - Pathology
    - Shaft is round in cross section with decreased diameter
  - Associations
    - May be seen with an epidermal nevus
    - May be a limited form of “uncombable hair syndrome”

Normal Strength

- Uncombable Hair Syndrome
  - Genetics
    - Autosomal dominant
    - Caused by premature keratinization of INNER root sheath
  - Clinical
    - Presents in early childhood; spontaneous improvement
    - Silvery-blond, “spun glass” appearance (angulated hair reflects light)
    - Disordered, can not be tamed!
  - Pathology
    - Nl in light microscopy; need scanning EM
    - Shaft is triangular in cross section, with a longitudinal groove (pili trianguli et canaliculi)
  - Associations
    - none

Course, ”spun-glass” hair

Note: tri shape and longitudinal groove

Normal Strength

- Loose Anagen Syndrome
  - Genetics
    - Unknown inheritance
  - Clinical
    - Blond girls, aged 2-5
    - Hairs are easily pulled from the scalp
  - Pathology
    - No external root sheath; premature keratinization of inner root sheath
    - Appearance of a “crumpled sock” on microscopy
    - >95% of scalp hairs in anagen
  - Associations
    - none

“Crumpled sock”

Normal Strength

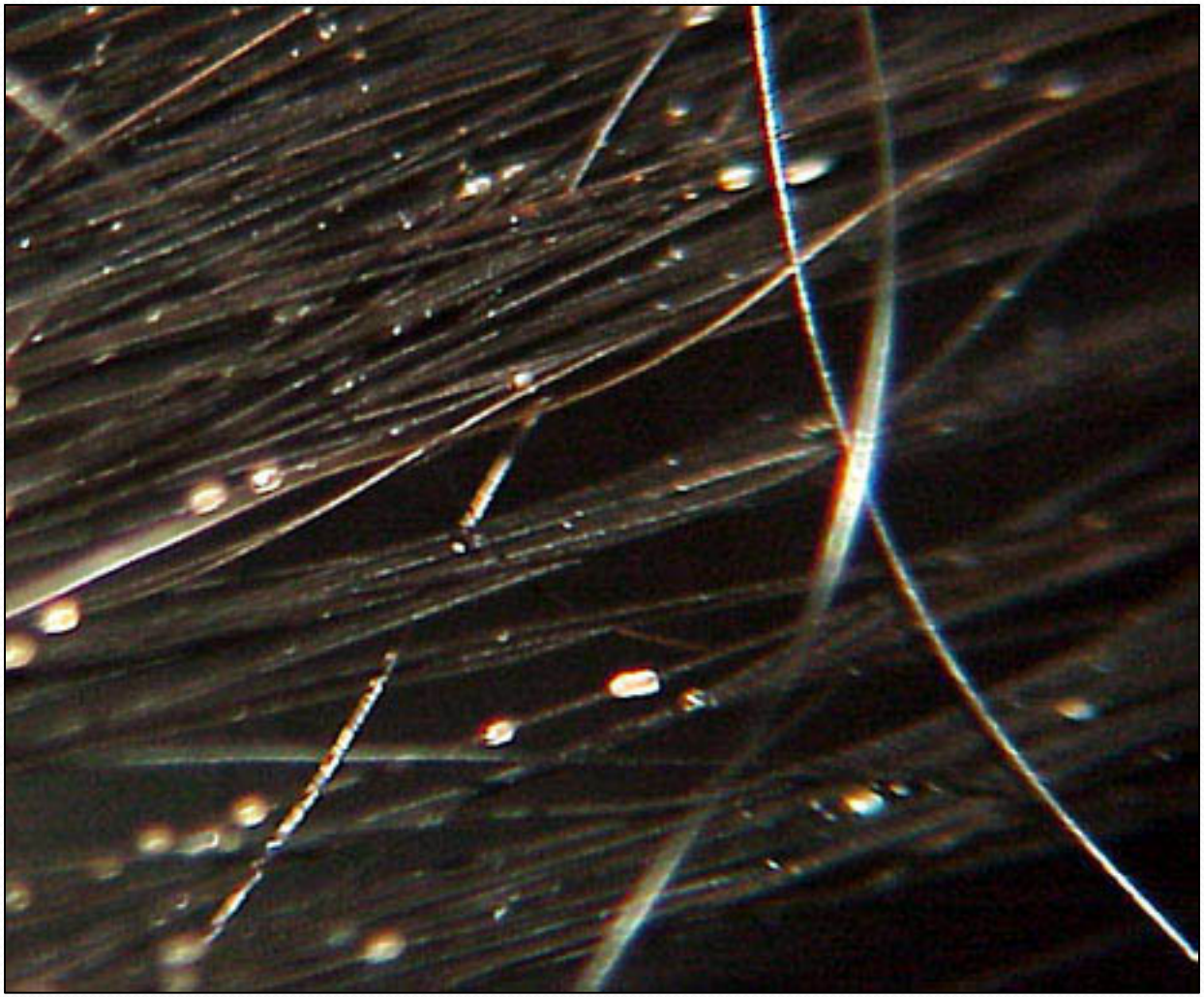
- Pili Multigemini/Bifurcati
  - Genetics
    - Unknown
    - Multiple papillae form hairs that exit ONE pilosebaceous unit
  - Clinical
    - Often unnoticed
    - Bifurcati: scalp margins in children
    - Multigemini: beard in adults
  - Pathology
    - Individual inner root sheaths, but a SHARED outer root sheath
  - Associations
    - Cleidocranial dysostosis

Trauma Induced

- Trichoptilosis
  - Split-ends
- Bubble Hair
  - Air bubbles within the hair shaft due to cosmetic damage (e.g. over-heating)
- Pohl-Pinkus constriction
  - Hair shaft constriction at time of metabolic challenge (e.g. low protein state, chemotherapy)
- Trichonodosis
  - “pretzel-like” knotting

Hair Shaft Deposits

- White Piedra
  - *Trichosporon beigelii*
  - Soft, white nodules; easily detached
  - Pubic>>scalp
  - Related to poor hygiene
  - Cut hair and use topical antifungals
- Black Piedra
  - Piedraia hortae
  - Gritty, black nodules on hair shaft
    - Penetrates shaft and then leaves concretions
  - Must cut hair to cure



White Piedra: soft spongy nodes all along hair shaft

Black Piedra: hard, difficult to remove, black concreations

Hair Shaft Deposits

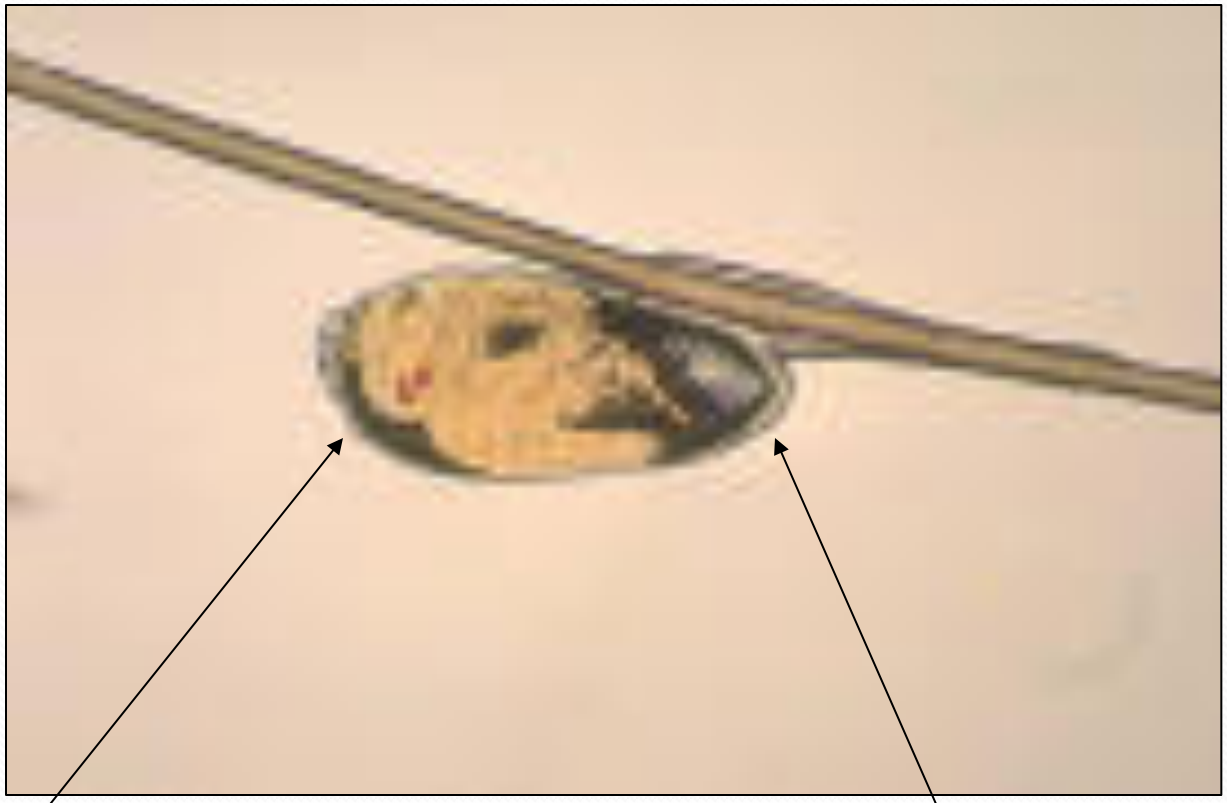
- Trichomycosis Axillaris
  - *Corynebacterium tenuis*
  - Superficial infection of the axillary or pubic hair
    - Yellow, black, or red concretions along hair shaft
    - Hairs may become brittle and break easily
  - Treat by cutting hair and applying topical clindamycin

Trichomycosis Axillaris

Hair Shaft Defects

- Nits: pediculosis capitis
  - Ovoid body attached by one end to the shaft
    - Operculum lies on “free” end
  - Near scalp
  - Difficult to move
- Pseud-nits: peripilar casts
  - Tubular casts that encircle the shaft
    - Usually from excess scale
  - Anywhere along shaft
  - Easily slide along

Pediculosis capitis: true nit



Operculum end, free to the air

Adherent end, encircling the shaft

White scale encircles
the hair shaft