Ten Cool Cases From Colorado:
Clinical-pathologic correlation and other puzzlers

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“Our job is to recognize the uncommon presentations of common disorders”
Case #1: Bump on eye

Recently adopted 2 year old Ethiopian female
HIV positive
“R/O cryptococcus”
Molluscum contagiosum
Molluscum contagiosum

Unique clinical presentations in immunocompromised patients

Clinical mimics:
- Cryptococcus
- Pyogenic granuloma
- Spitz nevi
- Juvenile xanthogranuloma
Case #2: Acne and warts

14 year old female

“Please treat resistant facial acne and periungual warts”
Angiofibromas and periungual fibromas
Tuberous Sclerosis

AKA: Bourneville Disease or Epiloia

(EPIlepsy, Low Intelligence, Angiofibromas)
Tuberous sclerosis

Autosomal dominant

Spontaneous mutation rate as high as 75%

Many clinical manifestations – varies patient to patient

Mutations in 2 genes:

- \textit{TSC1} (chromosome 9q34) - hamartin
- \textit{TSC2} (chromosome 16p13) - tuberin
TS – Major Diagnostic Criteria

Two or more of the following:

- **(L)** Lymphangiomatomatosis (lungs)
- **(A)** Angiomyolipoma, cysts (renal)
- **(R)** Retinal hamartomas (phakomas)
- **(A)** Angiofibromas or forehead plaque
- **(P)** Periungual fibromas
- **(S)** Shagreen patch-connective tissue nevus
- **(T)** Tubers (cortical)
- **(A)** Ash-leaf (hypomelanotic) macules >3
- **(R)** Rhabdomyoma (cardiac)
- **(S)** Subependymal nodules
TS - Minor Diagnostic Criteria

Dental enamel pits
Rectal polyps
Bone cysts
Cerebral white matter radial migration lines
Gingival fibromas
Non-renal hamartomas
Retinal achromic patch
Confetti skin lesions
Multiple renal cysts
Hypopigmented “ash leaf” macule
Forehead plaque
(Connective tissue nevus)
DEVELOPMENT OF CUTANEOUS FEATURES IN TUBEROUS SCLEROSIS

Percentage of affected individuals who have findings (%)

Age (years)

- Hypomelanotic macules
- Facial angiofibromas
- Shagreen patches
- Periungual fibromas

Case #3: Red plaque on face

Healthy 11 year old female
Asymptomatic plaque on face for about 4 weeks
“R/O discoid lupus”
Pityriasis rosea

Herald patch
Secondary lesions on neck arose about 1 week later
Often follow skin tension lines
Resolves spontaneously 8-12 weeks
+/- mild pruritus
If fever or lymphadenopathy, consider secondary syphilis
Patients should be in a gown!
Case #4: Liver disease and rash

20 mo old male, treated unsuccessfully for atopic dermatitis

Scalp with scale crust and discrete red papules, coalescing on bilateral post-auricular and parietal scalp

Small hemorrhagic crusted papules on penis and scrotum

“R/O seborrheic dermatitis”

Pertinent PMH:

Diagnosed 2 mo prior with sclerosing cholangitis, cause unknown
Reniform nuclei, with abundant lipidized cytoplasm
Langerin
Langerhans cell histiocytosis
Sclerosing cholangitis

Progressive course of cholestasis (decreased bile flow)
Inflammation and fibrosis of the intra- and extra-hepatic bile ducts
Chronic liver disease
### Sclerosing cholangitis

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<thead>
<tr>
<th>Primary</th>
<th>Secondary</th>
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<tbody>
<tr>
<td>Autoimmune</td>
<td>Autoimmune</td>
</tr>
<tr>
<td>Idiopathic</td>
<td>Immunodeficiency/AIDS</td>
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<tr>
<td>Inflammatory bowel disease (UC)</td>
<td>Intraductal stone/mechanical obstruction</td>
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<td>Adults</td>
<td>Abdominal trauma</td>
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<td></td>
<td>Intra-arterial chemotherapy</td>
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<td>Recurrent pancreatitis</td>
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<td>Eosinophilic cholangitis</td>
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<td>Systemic mastocytosis</td>
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<td>IgG4-SC (high IgG4, plasma cell infiltration)</td>
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**Histiocytosis**
Case #5: The linear rash

7 year old boy
3 week history of a linear rash on the cheek
Asymptomatic
He plays in the woods where there is poison ivy
“R/O contact dermatitis”
Lichen striatus

Sometimes the clinical history (from mom) is misleading
Uncommon, benign self-limited papulosquamous disorder
Asymptomatic (the reliable part of the history!)
Pathogenesis is unknown
Primary affects children, F>M
Most commonly affects extremities, rarely face
Most cases resolve in 3-12 months
Treatment = reassurance
Lichen Striatus
“I’m OK mom!!!”
Case #6: Pustular rash

9 year old male
2 day history of asymptomatic “rash” on trunk and extremities
No preceding illness
No previous history of skin disease
“R/O contact dermatitis, bug bites, infection, autoimmune”
Day 2
Day 5
Pustular psoriasis

Rare clinical presentation of psoriasis in children
+/- fever, arthralgias
May be triggered by URI, UTI, d/c of oral steroids, sunburn
HLA B27
HLA antigens B13, B17, Cw6 NOT seen
? Decrease in elafin (antileukoproteinase) → pmn infiltration into epidermis
DDX = AGEP, subcorneal pustular derm, SSSS, tinea
Case #7: Foot pain

17 year old female presents with intensely painful papules on feet
She is non-ambulatory because of pain
Has recently been skiing, and “bumps” started on last day of ski trip
“R/O exposure, infection, other”
Additional clinical history

A culture from the central pustule grew *Pseudomonas aeruginosa*

No organisms were visible with tissue gram stain or PAS

What recreational activity did she enjoy during her ski trip?
HOT TUB!
Pseudomonas Hot Foot Syndrome


Neutrophilic eccrine hidradenitis

Recreational exposure to *pseudomonas*

Exquisitely painful nodules on plantar feet

Self-limited disease

Other types of pediatric NEH:

- Chemotherapy-induced
- “Idiopathic”
Case # 8: Scary red blistering rash

“R/O Stevens-Johnson syndrome”

Lip biopsy pending
Staphylococcal Scalded Skin Syndrome

“But his skin culture is negative!”
SSSS: Blister is subCORNEAL
SSSS: Blister is subCORNEAL

SJS: Blister is subEPIDERMAL
SSSS (or pemphigus foliaceus!)
SSSS pearls

Remote staphyloccal infection causes toxin release → epidermal cell dyscohesion
  ◦ Skin culture negative

Flaccid superficial blisters
  ◦ Compare to SJS-TEN:
    ◦ Different blister plane
    ◦ No true mucosal lesions
    ◦ No targetoid lesions

Perioral involvement, crusting
“Sunburn” appearance, especially in groin/flexors
Skin pain, fussiness, discomfort
Stevens Johnson Syndrome

True target lesions with central epidermal change
Case #9: Purple hives
“Urticaria Multiforme”: A Case Series and Review of Acute Annular Urticarial Hypersensitivity Syndromes in Children

Kara N. Shah, MD, PhD, Paul J. Honig, MD, Albert C. Yan, MD
Urticaria multiforme pearls

Typical annular and polycyclic morphology of urticaria
Ecchymotic centers (“purple hives of Morelli”)
No target, mucosal, blistering, erosive lesions
Lesions last <24 hours
+/- low grade fever, mild leukocytosis and/or elevated APRs
+/- signs of illness (diarrhea, cough, etc)
Perioral, hand, foot edema without arthritis, arthralgias
+ Dermatographism
Antihistamine responsive
Urticaria multiforme vs. EM

Dusky center can mimic targetoid lesions of EM

Weston JA, Weston WL. *Pediatrics* 1992;89:802
Distinguish AAU from...

<table>
<thead>
<tr>
<th>ERYTHEMA MULTIFORME</th>
<th>SERUM SICKNESS-LIKE REACTION</th>
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<tr>
<td>TRUE TARGET LESIONS</td>
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<td>HANDS/FEET, NO EDEMA</td>
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<td>LESIONS NOT TRANSIENT</td>
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<tr>
<td>JOINT SWELLING/PAIN, FEVER, LAD</td>
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<td>LESIONS NOT TRANSIENT</td>
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<td>DRUG EXPOSURE</td>
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Cases #10a and 10b: Itchy red rash, getting worse
Patient #2
Diffuse tinea corporis

Patient #1: New kitten (Christmas gift)
Patient #2: Pet rats (her “best friends”)
Patient #1: Other family members affected
Patient #2: Only person affected

Both patients: Intense pruritus; poor response to antihistamines and topical/systemic steroids.

DDX in both cases: Psoriasis, SCLE, urticaria, pityriasis rosea. Prior biopsy was performed on patient #2 (“spongiotic dermatitis”)

Remember Microsporum species are relatively resistant to terbinafine
Thank you !