What's YOUR Diagnosis?

David O. Chastain, MD

DISCLAIMER
NEITHER THE PUBLISHER NOR THE AUTHORS ASSUME ANY LIABILITY FOR ANY INJURY AND OR DAMAGE TO PERSONS OR PROPERTY ARISING FROM THIS WEBSITE AND ITS CONTENT.

Disclosures

* BCBS of TN Regional Advisory Panel
12 yo WM presents to the clinic with an intermittent fever with rash, & knee pain x 3 wks

Systemic Onset JRA
(formerly called Still's Disease)

• 10-20% of all cases of JRA. Affects sexes equally.
• DX - combination of intermittent daily fevers >38.5°C (101.3°F) and arthritis. Fever must be present for at least 2 wks. Fevers are high and spiking and spontaneously return to NL on a daily basis.
• Difficult to diagnose because arthritis (necessary for the DX) may not be evident early in the disease.
• No lab findings are specific. Usually have high wbc with polys, thrombocytosis, anemia, and high ESR. ANA and RF are negative.
What medical condition must you consider when you find a bifid uvula?

Submucous Cleft Palate

• A close association exists between bifid uvula and submucous cleft palate.
• A submucous cleft can be diagnosed by noting a bluish line through the length of the soft palate.
• Affected kids have a 40% risk of developing persistent middle ear effusion.
• Many affected kids also have incomplete closure of the palate, resulting in hypernasal speech (exacerbated with adenoidectomy).

A 16 yo WF presents to the clinic c/o an enlarging, painful, pruritic rash on shoulder x 2 days.
Herpes Zoster (shingles)

- Herpes zoster (HZ), caused by reactivation of the varicella-zoster virus (VZV) that was acquired during a primary varicella infection.
- Characterized by dermatomal pain and a papular rash. The pain typically precedes the rash by several days and can persist for months after the rash resolves.
- PCR is the most sensitive and specific test for VZV DNA.

18 yo WF with cyanotic congenital heart disease (CHD)

Clubbing

- Pulmonary causes: lung cancer, lung abscess, empyema, cystic fibrosis, sarcoidosis, asbestosis, pulmonary TB
- Cardiovascular causes: congenital heart disease, infective endocarditis
- Endocrine causes: acromegaly, severe secondary hyperparathyroidism
- Nonpulmonary malignant causes: Hodgkin lymphoma, disseminated CML, thyroid cancer
- Gastrointestinal causes: UC, Crohn disease, celiac disease
- Other causes: cirrhosis, familial, pregnancy
15 yo WF presents to the clinic c/o an enlarging “lump” in her nose for 2 weeks that bleeds easily.

**Pyogenic Granuloma**
(granuloma telangiectaticum)

- Not caused by bacteria, as the name suggests, but is associated with capillary proliferation
- Often occur after a history of antecedent trauma
- Diagnosis based upon the clinical history of an erythematous, dome-shaped papule that bleeds easily and has developed over a few days to weeks
- Treatment is excision

13 yo OAF presents to the clinic with a 3-day history of fever, exudative pharyngitis, and a swollen tender neck.
Infectious Mononucleosis (Epstein-Barr virus infection)

- IM typically presents as fever, exudative pharyngitis, lymphadenopathy, hepatosplenomegaly, and atypical lymphocytosis.
- Subtle associated presentations are posterior cervical adenopathy, periorbital edema, and mildly elevated LFTs.
- EBV is associated with several other distinct disorders: X-linked lymphoproliferative (XLP) syndrome, post-transplantation lymphoproliferative disorders (PTLD), Burkitt lymphoma (Central Africa), nasopharyngeal carcinoma (SE Asia), and undifferentiated B- or T- lymphocyte lymphomas, Hodgkin & non-Hodgkin disease.

- Intermittent excretion in the saliva may be lifelong after infection.
- Incubation period is 30 to 50 days.
- Contact sports should be avoided until the patient is recovered fully from the IM and the spleen is no longer palpable.
- Because of potential adverse affects, corticosteroids should be considered ONLY for patients with marked tonsillar inflammation with impending airway obstruction, massive splenomegaly, myocarditis, hemolytic anemia, or hemophagocytic lymphohistiocytosis (HLH).

13 yo HM with a “blister that drains clear fluid when I bite it”
Mucocele

- Fluid-filled cavities with mucous glands lining the epithelium
- Typically seen after mild oral trauma, but they may also present on the labia
- Seen most frequently in patients younger than 20
- Vary in size and often contain a gelatinous fluid
- Spontaneous rupture can result in complete resolution, but removal is indicated if lesions are symptomatic.

15 yo HM presents with recurring spots in his mouth
Recurrent Aphthous Stomatitis (RAS) (canker sores)

- A common condition of the oral mucosa. Usually multiple, recurrent, small, shallow, round to ovoid ulcers with circumscribed margins.
- Etiology unclear.
- The RAS pt is afebrile, and has NO genital or ocular lesions, & NO previous Hx of immunodeficiency.
- Treatment is triamcinolone in Orabase.
- RAS has no underlying systemic conditions and is different from aphthous-like ulcerations (ALU).

Aphthous-like Ulceration (ALU)

- Oral ulcerations similar in clinical appearance to RAS can present as a manifestation in other diseases:
  - Behoet’s disease - recurrent oral & genital ulcers
  - Immunodeficiency - HIV
  - Vitamin/Mineral deficiencies - iron, folic acid, vit B12, zinc
  - GI problems - Crohn disease, celiac disease
  - Medications - NSAIDS, beta-blockers

12 yo WM presents to the clinic with a lump in his groin
Inguinal Hernia

- Diagnosis is clinical. Inguinal canal US and/or CT is helpful if positive, but a negative test does not exclude the diagnosis.
- Classified into Direct or Indirect (majority)

16 yo WF presents to the clinic for a pelvic exam
16 yo WF c/o worsening sores on hands x 2 wks

**Impetigo**

- *Staphylococcus aureus* (MRSA or MSSA), *Streptococcus pyogenes*, or a combination of the two organisms
- Bullous impetigo is due to *Staphylococcus aureus*.
- Suspicion of MRSA is raised in cases of spontaneous abscess or cellulitis.
17 yo WM presents to clinic c/o a rash 1 wk after being treated with ampicillin for a sore throat that hasn’t gone away.

**Ampicillin-induced Rash of EBV**

- Develops in many older adolescents 5-10 days after starting treatment with ampicillin, amoxicillin, or beta-lactam antibiotics. Even though it is associated with penicillins, this rash does NOT represent a true penicillin allergy.
- Rash is typically maculopapular and pruritic.
- Rash resolves in a few days after discontinuing the antibiotic.

17 yo BM comes to clinic for “bumps on my package.”
Herpes Simplex Virus (HPV)

- HSV-1 or HSV-2 can cause oral, genital, and ocular lesions. Most people have unrecognized disease.
- Symptoms range from asymptomatic to recurrent painful ulcerations.
- HSV PCR of active lesions is more sensitive than HSV viral culture.
- Genital HSV-2 infection is more likely to recur than is genital HSV-1 infection.
- Acyclovir, famciclovir, and valacyclovir are effective at shortening the duration and severity of an outbreak.

16 yo WM comes to clinic c/o recurrent itchy rash for several months
Urticaria (hives)

- Urticaria are erythematous, blanching, edematous, nonpainful, pruritic lesions that typically resolve within 24 hrs and leave no residual markings.
- Acute urticaria lasts <6wks and is generally self-limited. Chronic urticaria last >6wks and is complex.
- Angioedema involving the face or neck requires prompt treatment with epinephrine.
- H1-receptor antagonists (antihistamines) are the mainstay of treatment for acute & chronic urticaria.

14 yo WF who c/o “weird-looking” feet

Hallux Valgus Deformity (bunion)

- Valgus malformation of the great toe is a very common and potentially painful and debilitating condition of unclear etiology.
- The deformity is a lateral deviation of the great toe (hallux) on the first metatarsal.
- The prevalence is greater in shoe-wearing populations than barefoot populations. Also more common in women than men.
13 yo HM who c/o an increasing number of freckles

Neurofibromatosis type 1 (NF-1)

- 1 in 4000 people with no gender, race, ethnic, or geographic distribution
- Autosomal dominant but in half of people affected, the condition results from a new mutation
- Café-au-lait spots increase in size and number with age, especially at puberty and with pregnancy
- Lisch nodules are iris nevi visible only with slit lamp exam and are present in almost all NF-1 individuals by 20 years of age

NF-1 Diagnosis
(2 or more of the following must be present)

- >6 café-au-lait spots at least 5mm in diameter in children and 15mm in diameter in adults
- 2 or more neurofibromas of any type or one plexiform neurofibroma
- Axillary or inguinal freckling
- 2 or more Lisch nodules
- Distinctive osseous lesions such as sphenoid dysplasia or thinning of long bones with or w/o pseudarthrosis
- Optic glioma
- A first-degree relative with NF-1 (based on criteria)
16 yo BM c/o recurring sores under his arms

Hidradenitis Suppurativa (HS)
- A chronic follicular occlusive disease involving the intertriginous skin of the axillary, groin, perineal, and inframammary regions.
- Clinical course is variable. Onset is at puberty.
- The etiology and pathogenesis of HS is unknown. The significance of bacterial infection in HS is controversial.
- Predisposing factors: obesity, endocrine (sex hormones), genetic, and possibly smoking, lithium, & depo.

Hidradenitis Suppurativa (HS)
- The usual presentation is of small painful subcutaneous nodules. Nodules develop and may rupture, discharging purulent, sometimes malodorous material and forming sinus tracts.
- The diagnosis is based upon characteristic clinical manifestations.
- Treatment depends on severity. I&D often leads to recurrence and antibiotics may need to be used for 6-8 wks at a time. Plastic surgery may ultimately be needed.
16 yo WF with an enlarging lump in her neck x 3 wks

You suspect the mass involves the thyroid. How do you want to evaluate it?
16 yo WM c/o an itchy rash on his hands and penis for two weeks

The CDC recommends treatment with:

• A) Permethrin 1% lotion
• B) Lindane 1%
• C) Permethrin 5% cream
• D) Oral Ivermectin

Scabies

• Caused by infestation with sarcoptes scabiei, a microscopic mite that burrows and deposits eggs and feces in human stratum corneum
• Transmission usually occurs via direct skin-to-skin contact.
• Crusted scabies (scabies crusted, Norwegian scabies) is a distinctive and highly contagious form of scabies that typically occurs in pts with a history of immunosuppression, neurologic disorders or institutionalization.
Scabies

- Pts with scabies invariably complain of pruritus, particularly at night.
- Humans are the source of the infestation. Infestation from animals is uncommon, and animal mites do not replicate in humans.
- Household members should receive prophylactic therapy.
- Special attention should be given to trimming the fingernails and applying medication to these areas.
- Mites do not survive more than 3 days without skin contact.

15 yo WF presents to the clinic c/o “funny fingers”

Clinodactyly

- Curving of the little finger toward the ring finger
- A minor congenital malformation. The middle bone of the 5th finger is underdeveloped and, instead of being rectangular, is wedge-shaped.
- Occurs as an isolated finding in normal people or in association with other congenital malformations. It is a common component of Trisomy 21 (Down syndrome) and Klinefelter (XXX) syndrome.
- No treatment is required.
16 yo HM presents to the ED with painful hands and thighs
Suicide Attempt by Mercury Injection