Funny Hoof Beats or Zebras? Diagnostic Dilemma Cases in a Primary Care Office

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Disclosures

• No relevant financial disclosures.
Objectives

• Utilize case-based learning to expand our differential diagnosis skills.
• Develop skills in ordering and interpreting less-frequently used laboratory tests.
• Learn from “diagnostic clinic” patients to diagnose similar patients in primary care.
• Appreciate how a “diagnostic dilemma clinic” can help in the care of our pediatric patients.
Objectives

• There are separate “learning points” at end of each case—for a given percentage of you, these are obvious and you know them—please just be patient 😊
• We’ve all been there...
  – New patient, nine months of vague symptoms. No context since you’ve never seen them...
  – Parents understandably anxious.
    • That drives them to want an answer, any answer, fast answer.
  – We feel anxious.
    • Slow, thoughtful work-up better than shotgun work-up.
    • Patient is neither going to die nor get fixed overnight.
      – But the family sees it differently
  – This dynamic sets us up for failure.
• These are the types of cases we’ll mostly tackle today.
  
  – We’ll talk about strategies that can translate from “diagnostic clinic” to your primary care office
What IS Diagnostic Clinic, anyway?

• Glad you asked!
• “Opened” in fall, 2011, as an entry point to get a thorough evaluation at Duke for children whose symptoms don’t neatly fall within a given subspecialty.
• Comprised of me and Dr. Richard Chung
  – Dr. Chung sees most of the adolescents 😊
• Other academic centers have them ,too!
Referral Sources

• Community primary care physicians
• Pediatric subspecialists
• Duke Children’s Primary Care
• Duke Peds ED
• Friend of a friend of a friend of a friend...
  – Almost never ends well
Types of Cases/Referrals

- True diagnostic dilemma, unclear where to start.
- Pretty complete work-up done, PCP not sure if missing anything.
- Parental reassurance even though referring provider pretty sure that patient is okay.
- *Referring provider sure that patient is okay but is worried about a factitious disorder (MBP)*
Disclaimer that I tell patients

• I spend 99% of my clinical time seeing pediatric primary care patients in 10-15 minute slots.

• When I get referrals, I take several hours to review chart and read.
Disclaimer that I tell patients

• In other words, there’s nothing wrong with the referring provider if 1) they don’t figure out a diagnosis and 2) they don’t spend an hour with their patients

• I also have easy email access to a ton of specialists= free lifelines!!!
So...

• With that being said, some of the things I’m going to suggest may be easier said than done.
Some scripting

• “We are going to do what we need to do to evaluate your child.”
• “Sometimes we find an answer, often we do not.”
• “Sometimes children’s bodies are smarter than the tests we have to diagnose them.”
  – “But we will make sure we don’t miss anything dangerous.”
Bizarre Analogy

- “Even if we do not find an underlying cause or disease, we will still work to make them feel better and get them back to the ‘old Johnny’.”
• Enough philosophy.
• Time to do some patient care!
Case #1

- 10 y/o female with right elbow pain and swelling for past seven months.
- At time of onset, she had fever, sore throat, positive rapid strep.
HPI

• Symptoms resolved after one week of regular ibuprofen.
• Yay, we’re done!!!!!
• After 4-6 weeks, though, developed swollen and painful left wrist.
• This, too, improved after one week of ibuprofen.
HPI

• 3 months later, developed fever/sore throat/diarrhea concomitantly with pain in both elbows as well as left knee/hip/wrist.

• Given concern for dehydration and degree of illness, seen in ED
  – Had bump in inflammatory markers, noted by PCP
    • Diagnosed w/ viral syndrome, given symptoms, causing a flare of her background arthritis.
PCP

- Astutely realized that something NQR...
- NOT
- QUITE
- RIGHT
What was “NQR”? 

- **Multiple discrete episodes of joint pain/swelling**, none of which associated with trauma.
- **Different large joints involved** *(we’re now up to five, by my count).*
- **Objective evidence of inflammation** by labs outside of what a viral process should do.
PCP’s Next Steps

• 1) Repeat labs two weeks later.
   – Normal white count at 7.9, platelets inc’d at 577, CRP down to 2.6 mg/dL, sedimentation rate down to 59.

• 2) Refer to peds rheumatology
   – ~ 1 week after repeat labs, 3 weeks after ED
     - ESR 56, and CRP 0.85. ASO was 161, Anti-DNAse B normal at 100. Platelets 507, elevated AST/ALT
Additionally

– CMV IgM negative, parvo IgM negative, parvo IgG positive, and EBV titers as follows:
  • Positive EBNA, VCA IgG, and Early Antigen IgG with negative VCA IgM).

– Diagnosed with reactive arthritis in setting of likely EBV infection.

– Advised to f/u labs serially.
EBV Review

- EBNA
- EBV IgM
- EBV IgG
- EBV early antigen
Positive EBNA, VCA IgG, and EA IgG
Negative VCA IgM
Next Month

• Asymptomatic
BUT...

• Two weeks later:
  – Recurrence of left knee pain and swelling accompanying malaise.
• Referred to diagnostic clinic for further work-up.
Referral Question

• Source of patient’s recurrent joint pains?
  – Specifically, why are these getting worse even though EBV infection should be resolving?
  – Makes no sense. Shouldn’t they just see peds rheum again???
ROS

• No vomiting
• Occasional abdominal pain- occurs when has diarrhea, required a few ED visits
• No jaundice
• No cold sores or other oral lesions
• No rash
• Occasional fevers
• No weight loss, but no weight gain x7 months
• Pre-menarcheal
Social History

- Lives with mom, dad, 4 siblings.
- No smokers at home.
- Goes to 5th grade, does well, hasn't missed more than a couple of days of school. Enjoys school.
Family History

- Mom - HTN
- Dad - healthy other than benign proteinuria
- MGF - Crohn's disease, HTN
- MGM - type II DM, HTN
- PGF - deceased, unsure cause/age
- PGM - diabetes mellitus (mom unsure which type), HTN
- 3 brothers, 1 sister - all healthy other than allergic rhinitis
Physical Exam

- **Gen:** Well-appearing, NAD, pleasant.
- **HEENT:** MMM, TMs w/o effusion, no oral ulcers, good dentition, no lymphadenopathy, no thyromegaly. *Conjunctivae slightly pale.* No scleral icterus.
- **CV:** regular rhythm, no murmur, 2+ pulses.
- **Resp:** Normal WOB, lungs clear bilaterally.
Physical Exam

- Abd: soft, non-tender, non-distended, no organomegaly, normal bowel sounds
- Skin: No rash
- Ext: no edema, FROM all joints, no edema/warmth of joints
- G/U: Tanner II pubic hair, tanner II breast buds
- MSK: 5/5 strength, no scoliosis
Problem List

• Recurrent joint pain/swelling
  – Poly-articular arthritis
• Elevated inflammatory markers
• Likely EBV infection leading to elevated liver enzymes
• Exam normal other than conjunctival pallor
Hmm, I’m stuck

- I asked the patient what concerned HER the most.
Patient’s concerns, when asked:

• Joint swelling
• Diarrhea
  – Some during that illness precipitating ED visit but sporadically since.
  – Even associated with blood on 3 different occasions.
• Difficulty swallowing for one week earlier in course - improved with ranitidine.
Problem List - Original

• Recurrent joint pain/swelling
  – Poly-articular arthritis
• Elevated inflammatory markers
• Likely EBV infection leading to elevated liver enzymes
Problem List- Modified

• Episodic arthritis
• Elevated liver enzymes
• Diarrhea, tenesmus
• 3 episodes bloody stool
• Exam normal other than conjunctival pallor
  – Note: these are all observations and symptoms, not diagnoses.
Problem List- Modified

- 3 episodes bloody stool
- Elevated liver enzymes
- Episodic arthritis
- Diarrhea, tenesmus
- Exam normal other than conjunctival pallor
  - Note: these are all observations and symptoms, not diagnoses.
So...

• The question isn’t: “What’s causing this child’s arthritis?”
• The question should be: “What’s wrong with this child?”
Differential of the arthritis?

- **Infectious/reactive:**
  - Lyme
  - rheumatic fever (unlikely);
  - post-strep reactive arthritis
  - viral reactive arthritis (EBV, CMV, HIV, hepatitis, HSV)
  - *Yersinia* intestinal infection.

- **Rheumatologic:** JIA, reactive arthritis

- **GI:** Crohn's Disease, Ulcerative Colitis, Wilson's Disease
Diff dx-Taking all into account

• IBD
• IBD
• IBD
• IBD
• IBD
• Yersinia
Oh yeah...

- What WAS that family history again?
Family History

• Mom- HTN
• Dad- healthy other than benign proteinuria
• MGF- Crohn's disease, HTN
• MGM- type II DM, HTN
• PGF- deceased, unsure cause/age
• PGM- diabetes mellitus (mom unsure which type), HTN
• 3 brothers, 1 sister- all healthy other than allergic rhinitis
Diagnosis?

- IBD, likely Crohn’s disease.
Further Care

• Discussed w/ Peds GI- did not recommend IBD panel; they saw patient briefly thereafter (~1 month).

• Would’ve been even faster (~1 week), but family was traveling.
Endoscopy-Gross and Histopathology

Consistent with Crohn’s Disease and Autoimmune Hepatitis

Stomach
Terminal Ileum
Rectum
Lag-Time Common

• Many children with IBD have long lag-time of symptoms before diagnosis (Kwon, YH, and Kim, YJ, 2013).
More re: Lag Time

- 2.5 months for arthritis preceding diagnosis
- 5.8 months for abd pain
- 4 months for diarrhea
- 4.1 months for hematochezia
- 3.8 months for weight loss
- (Heikenen et al 1999).
  - Growth failure- 18 months.
Aftermath

• Did well for awhile, but had persistent liver dysfunction refractory to steroids and 6MP.
  — Subsequently diagnosed with mixed sclerosing cholangitis and autoimmune hepatitis

• Now on 6MP, prednisone, Humira but energy level and appetite normal, attending school.
Key Points

• Seeing entire chart is important.
  – Leverage your EMR

• If asked family history as “any family history of joint pain/arthritis”, answer would be “no.”

• Asking patient what HER symptoms were led to the diagnosis

• Making problem list based on what was known, not what seemed to be known.
That said...

• Kind of helped to have three hours to read through chart, make timeline, make problem list, AND an hour with the patient.
Learning Points

• Include IBD on your differential for joint pain, especially if associated GI labs/symptoms
• Include IBD in differential early for abdominal pain, given long lag-time before diagnosis
• IBD panel won’t be helpful in diagnosis
Case #2- “The Truth?”

• Challenge of this case: figuring out what info passed along in the referral is true, what is not true, and what is merely true-true unrelated.

• Not sure how all the given subspecialists were supposed to help this kid—through nobody’s fault, all were led down a winding path.
Case #2

- 11 y/o male with ~2 years of left hip pain
  - Now has more recently had:
    - 5 months prior to appt- wrist pain
    - 1 month prior- neck pain after MVC; also developed swollen left knee and right ankle
Referral Questions

• Why is he having hip pain?
• If at all, do his more recent symptoms relate to his 2 years of hip pain?
More HPI

- Right ankle pain is currently 10/10.
- Pain is no longer improved by NSAID’s.
- Has struggled to bear weight, although this has improved in recent days.
Previous Hip Evals

• Seen by ortho, heme-onc, rheumatology over course of first year of symptoms.
• 6 mo ago- MRI-signal enhancement of left SI joint and greater trochanter, right acetabulum.
• 5 mo ago- bone scan-concern for eosinophilic granuloma.
  • Bone biopsy done- negative
• 2 months ago- CT with arthritic changes in both hips.
Function

- Has intermittently in past used a walker and wheelchair when his “hip locks”
Acute on Chronic Illness

• Even zebras get sore throats!
• Presented to ED with fever/sore throat, presumed strep
  • Given joint pain history, bloodwork done, ASO positive. Diagnosed w/ strep, advised to f/u with PCP to discuss possible rheumatic fever.
• Treated w/ PO keflex and then IM Penicillin-started earlier in month for presumed rheumatic fever (more on this later)
ROS

- Loose stools since infancy - non-bloody
- No vomiting, no abdominal pain
- Transient appetite - ? weight loss
- No fevers; occas. night sweats; no swollen LN’s
- No changes in vision
- No systemic rash - just dry skin on ankle
- + sternal chest pain, no SOB
- No change in urination recently (Had incontinence several months ago)
- No back pain
Prior Work-Up

• Per report, negative cardiology evaluation
• Elevated platelets (400’s)
• HLA B-27 +
• Negative Lyme, negative ANA, negative RMSF.
• ESR 24 (down from 29 earlier)
• CRP increased from 0.35 mg/dl two months ago to 7.1 last month
• ASO titers 412
Past Medical History

• Autism Spectrum Disorder
  – “High-functioning”
• Chromosome 3q29 deletion
Family History

- Non-contributory
Social History

• Lives with mom and 3 brothers.
• In 5\textsuperscript{th} grade in “special classroom”
• Father not involved
The Questions

• Is this rheumatic fever vs post-strep arthritis?
• Another cause?
Physical Exam

- Weight: 48.7 kg (87\textsuperscript{th}%); Height 150 cm (69\textsuperscript{th}%); BMI 21.644 (90\textsuperscript{th}%)
- Weight 6 months ago (51.8 kg)
- Temp 36.3, P 95, R 24, bp 110/68 (62\textsuperscript{nd}/68\textsuperscript{th})
- General: well-appearing, talkative, utilizing walker on arrival to clinic
Physical Exam

- Neck: Neck supple. No adenopathy or thyromegaly.
- CV: Regular rate and rhythm, no murmurs, rubs or gallops. Distal pulses 2+ & symmetric. Capillary refill < 2 seconds. Note normal blood pressure.
- Lungs: Clear to auscultation bilaterally; no wheezes, rhonchi, or rales.
Physical Exam

• Abd: Soft, non-tender, non-distended. Bowel sounds present. No hepatomegaly or masses. **Spleen tip palpable 1 cm.**
• GU: tanner I male, testes descended bilaterally.
• Skin: **six petechiae on abdomen; dry skin on lateral surface of left ankle.**
• Neuro: Alert, active.
Physical Exam-Ext and MSK

• Both knees without effusions/swelling.

• Right knee tender with ballottement of patella. Left knee non-tender. FROM both knees.

• Dec'd ROM of flexion of left hip, right hip wnl.

• Right ankle warm with obvious edema and marked tenderness. Left ankle slightly warm/red, mildly swollen and tender.
MSK exam (cont)

- Tenderness bilateral SI joints.
- All upper extremity joints with FROM and no edema/ tenderness. Toes without edema/tenderness.
- Normal bulk & tone.
- Able to bear weight and ambulate. Negative Schober test.
Problem List

- Hip pain, transient ankle/knee pain
- Multiple abnormal joints (~6) on exam
- Recent strep infection
- Chromosomal abnormality
- Weight loss
Key Point

• No actual evidence of a strep infection.
• Need either rapid strep OR positive throat culture OR elevated ASO titers that improve dramatically with antibiotics.
  – Thus, we’ll need to repeat ASO titers
    • Improved from 412 to 332 (not considered dramatic improvement).
• Additionally, keep in mind that most symptoms pre-dated the ? strep illness.
Ancillary Tests

- ecg- pr interval normal (124)
- u/a normal, normal protein:creatinine ratio
- CMP normal
- Normal WBC and differential
- Borderline anemia- hct 34, not microcytic
- Platelets elevated at 484
- ESR 60
- CRP 0.48 mg/dl
- HLA B-27 positive
Revised Problem List

- Multi-joint arthritis, non-migratory
- Strep exposure but probably not recently
- HLA B-27 positive
- Elevated inflammatory markers
- Weight loss
- Mild anemia
Possibilities

- Acute rheumatic fever?
- Post-streptococcal arthritis?
- Polyarticular JIA?
- Pauciarticular JIA?
- Enthesitis-related arthritis?
Diagnosis of ARF

- Group A infection followed by 2 major or 1 major and 2 minor

**Major:**
- Migratory arthritis (predominantly involving the large joints)
- Carditis and valvulitis (eg, pancarditis)
- Central nervous system involvement (eg, Sydenham chorea)
- Erythema marginatum
- Subcutaneous nodules

**Minor:**
- Arthralgia
- Fever
- Elevated acute phase reactants (erythrocyte sedimentation rate [ESR], C-reactive protein [CRP])
- Prolonged PR interval
Post-Strep Reactive Arthritis

• Latent period between the antecedent streptococcal infection and the onset of migratory arthritis is shorter (one to two weeks) than seen in classic ARF.
• The response of the arthritis to aspirin/NSAID’s is poor in comparison to the dramatic response seen in classic ARF.
• Carditis is not seen in these patients.
• Severity of arthritis is quite marked.
• Renal abnormalities often are seen in these patients.
• Acute phase reactants tend to be lower than in the setting of ARF.
Polyarticular JIA

• By definition, 5 or more joints in first 6 months.
Pauci-articular Arthritis

- Rare to present after pre-school age, especially rare after 10 y/o
- Almost never begins in hips
- Inflammatory markers typically negative
- ANA typically positive
Enthesitis-Related Arthritis

- Children with arthritis and enthesitis, or arthritis and two or more of the following:
  - Sacroiliac joint tenderness
  - Inflammatory spinal pain
  - HLA-B27
  - Positive family history of anterior uveitis with pain, a spondyloarthropathy, or inflammatory bowel disease
  - Anterior uveitis associated with pain, redness, or photophobia
Enthesitis-Related Arthritis

• Children with arthritis and enthesitis, or arthritis and two or more of the following:
  – *Sacroiliac joint tenderness*
  – Inflammatory spinal pain
  – *HLA-B27*
  – Positive family history of anterior uveitis with pain, a spondyloarthropathy, or inflammatory bowel disease
  – Anterior uveitis associated with pain, redness, or photophobia
Our Patient

• Enthesitis-related arthritis
• Course: started on piroxicam
• Referred to peds rheum at Duke
  – Given lack of successful response, treated with Enbrel, sulfasalazine, joint injections
What did I learn? (and take-home points if you didn’t know these)

• ASO titers normally elevated in school-age children.
• Poly-articular arthritis requires 5 joints in first 6 months of illness.
• This was rare instance where checking ANA is useful (negative test gives a lot of info).
What else can be learned?

- As you take over the primary care of a patient that just moved here, don’t assume that diagnoses attached to a patient are automatically correct.
  - Within the scope of your practice, take a moment to think about how you can address this
- Utilizing time lines can be REALLY helpful.
What else can be learned?

– Elevated ASO was “true,” past exposure to strep was “true,” arthritis was “true.”
  • But relationship between strep and arthritis was not causal.
Case #3

• Friend of a friend type referral.
  – Adult rheumatologist emailed peds rheumatologist about a family friend (the mother previously worked in his office)

• Uh oh!
Per the email

- 13 yo girl with a history of being injured in a soccer game with a possible concussion and months later developing weakness
HPI

- Pretty healthy for first 12 years
- May- frequent headaches.
  - In context of "severe viral illness"
    - LP negative for meningitis
    - head CT that was normal
    - “relatively normal” ophtho eval (? Concern for optic neuritis)
- July -concussion at soccer camp-had negative CT scan, improved after a week
HPI- cont’d

• A week later:
  – abdominal bloating after eating, nausea/vomiting.
  – Had endoscopy and colonoscopy done with biopsies and everything came back normal.
  – Then had elimination diet without change in symptoms (rice, chicken, water).
The Fun Begins

• Right eyelid drooping on 10/3 that progressed over the day to right sided facial drooping.
• She was seen at her PCP that afternoon, and told it was likely Bells Palsy, and given prednisone. Lyme disease testing was done
• Weakness progressed over the following day, and again called her PCP, who gave reassurance.
• Pts family decided to drive to Duke ED for further evaluation. At that time, she had a normal brain MRI, and was given a course of valcyclovir, discharged home
Next

- On 10/5, pts mother heard her yelling, and found her face down on the floor of her room. Pt states that her "legs went out" and she fell.
- Denied LOC or any injury during the fall.
- She was brought to local ED, where workup included normal head CT, CXR, CBC, glucose, CMP, PTT, negative UDS, EKG with sinus bradycardia.
- Admitted for observation.
Other changes

• Developed RUE DVT
  – OCP plus immobilization?
• Vision went from 20/15 or 20/20 to 20/60 in right eye.
• Reports blurry vision right eye only.
• Saw ophthalmologist after concussion but this preceded vision change.
• Recent flu vaccine
Visit with me-

- Nothing new to HPI
Social Hx

- Lives with mom, stepdad, dog, cat (Adopted by stepdad in 2010)
- Likes soccer, gymnastics, surfing
- Bio dad in MVC when she was 3 y/o, had TBI.
- 8th grade, attends private school. Expelled from previous school in 6th grade after a prank
- Life is 6 out of 10. “I love life”. "wish I wasn't in hospital". NOT stressed, anxious, nor depressed.
More Social Hx

- Life previously 8 out 10. Biggest stress used to be how to wear her hair.
- Has never smoked/drank.
- Has never thought about self harm.
- Attracted to boys. Never been sexually active.
- Uncle passed away from liver cancer in July.
- Stepdad's work partner had several deaths in family (someone fell in family broke neck, etc).
Exam

• In wheelchair, right arm resting on armboard and right leg extended, wearing ankle brace
• Quiet, slightly flat affect
• normal bulk 4/5 strength LUE and LLE, 0/5 strength RUE and RLE
• Perrl, eomi, fundi normal. Tongue deviates to left when protruding, midline at first opening mouth before protrusion.
• 2+ patellar reflexes bilaterally.
• Absent sensation to touch on right arm and leg.
More exam

• Sensation stops at distal third of clavicle on right side of chest and on back.
• Touch intact on forehead, not on right side of face except just at lateral surface of nose.
• Absent proprioception on right, left normal.
What’s going on???

• 13 y/o female with Bell’s palsy and subsequent right sided weakness and anesthesia. Reflexes intact.

• “find the lesion” from neuroanatomy class.
Weakness/anesthesia

- Stroke
- Tick paralysis
- Toxins
- Myasthenia gravis
- Porphyria
- Guillain-Barré
- Transverse myelitis

- Polio
- Spinal cord tumor
- AHC
- Stroke
- Hemorrhage
- Conversion
Exam at neurology

- MS: Flat affect initially, tearful when discussing dx, stressors.
- Face symmetric at rest (normal NLF)-decreased excursion of R face with smile, both eyebrows raise, tongue midline at rest and with speech but deviates slightly to L with protrusion. Facial sensation mildly decreased on R.
- Hearing grossly symmetric.
• Motor: Normal tone, bulk and 5/5 strength throughout On L. R with low-normal tone and no movement on testing or with noxious stimuli. No synergistic or counter-pressure on opposite side (hoovers sign).

• Sensation: Intact on L. No reported sensation to light touch, pin scratch on R extremities or trunk to midline.

• Reflexes: Present and symmetric in upper and lower extremities with no clonus or spread. Totally normal reflexes.
• Gait: Stands from chair with 1 assist. Stands alone with 4 prong cane in L hand. Mom drags R leg forward by pant leg, then steps with L hand and cane. Can step with the left with no additional assist than the cane.
More Social Hx (at neuro)

• Apparently stepfather tried to “buy out” parental rights from bio father.
• Bio dad more recently trying to get more involved.
• Steppadad having trouble coping due to own h/o parental abandonment and h/o PTSD from military.
So yes...

- This poor kid has conversion disorder.
- Remember, child is NOT faking nor trying for secondary gain.
- She’s actually quite distressed by her symptoms.
Treatment Contingent on Buy-in

• So don’t throw the term around loosely.
  • Be thorough & intentional before stating this (and thorough does NOT have to mean million dollar workup)

• Explicitly explain what you’re thinking and why.

• Don’t use the word “faking”.

• Don’t do more work-up once you have diagnosis.

• Do see them in clinic for recurrent symptoms.

• Do let them know you won’t abandon them.

• Therapy only effective once patient agrees w/dx.
Case #4

• Very different type of case
  – But shows how a diagnostic clinic can be an effective portal into tertiary care
  – And if nothing else, how important it can be to advocate for your patients even if you don’t know what their underlying problem is
Case #4

- 4 month-old referred from PCP for history of VSD, periorbital edema, and ?petechial rash.
  - AGA term infant. Only complications: AMA and GDM.
  - Growing great.
  - Seems “tired,” less active than older siblings did.
- My exam notable for hypotonia, telangiectatic rash, nystagmus.
  - TORCH vs genetic vs neuromuscular disorder.
Example of why model can help

- Referred on 2/18
- I saw patient on 2/27
  - Obtained labs
  - Ordered head u/s, renal u/s, both done next week
  - Sent copy of evaluation to PCP that evening
  - Suggested that PCP get CDSA involved- eval set up by mid March
Time Course

• Seen by ophtho and neuro 3/23
  – Found to have optic nerve atrophy
  – Further lab work-up done, including elevated AFP
  – AT gene and SMA gene testing negative
• ENT 3/27- laryngomalacia but cleared for MRI
• Genetics consult 3/31
• MRI 5/16-delayed myelination and decreased periventricular white matter, enophthalmos, sphenoid wing dysplasia
• 5/21 – genetics follow-up
Diagnosis and Subsequent Care

• 7/6- GM1 Gangliosidosis
  – Unfortunately, devastating diagnosis.
• Enrolled in hospice by September.
• Followed by GI, neuro, ophtho, ENT, cardiology, genetics, SLP
• Passed away the following July (1 year after diagnosis, at 21 months of age)
For this kiddo

• I couldn’t make diagnosis myself.
• The immunoglobulin profile not the best test to look at A-T given age of patient.
• I could grease wheels to get consults done quickly.
  – How can you do this?
• Child plugged in for developmental services while awaiting diagnosis.
• Able to provide answer for family
  – Mormon family in small community
  – Recurrence risk an important concern
Other Recent Cases

• Some quick fun (or not so fun) ones.
Fallen Thru Cracks

• 11 y/o with history of Ehlers-Danlos diagnosed elsewhere, now with recurrent abdominal pain.
  – Helped family to know that abdominal pain is common in EDS.
  – Ensured they got plugged in to cardiology
Recurrent Dizziness

• Initially a “sigh” referral.
• 14 y/o male with h/o dizziness, previous admission at Wake Med, diagnosed w/ conversion disorder.
  – Family accepted diagnosis
• New recurrence of chest pain with dizziness, referred to me.
• Convincing enough that I sent to cardiology.
  – Delta waves on ECG, diagnosed w/ WPW.
  – Ablated, now asymptomatic
Other Recent Cases

• Two siblings with extensive work-ups.
  – Frequent change of PCP.
  – Frequent seeking of work-up.
  – “Virtual” geneticist diagnosed with mitochondrial disorder, ? MELAS
  – Both with ports for IVF for POTS. On carnitine and arginine supplementation.
  – Children normal on exam.
  – No abnormal labs. Ever.
Social Factors

- Mom and dad divorced after dad returned from Afghanistan.
- Mom trained as nurse.
Siblings

• Diagnosis: medical child abuse.
• Bringing to follow up in clinic to go through a care plan/contract.
• Suggest mental health care.
Take-Home Points

• These diseases didn’t form overnight.
  – Ok to not solve them overnight

• Most kids don’t have rare diseases.
  – More common to have common disease presenting unusually.

• For the kids that do have unusual presentations, consider a diagnostic clinic-type referral if you don’t know where to start.
Formal Referral Process

• Discuss with your patient!
• Call 1-888-ASK DUKE and mention you’d like to refer to peds diagnostic clinic.
• Referral center will fax your office a questionnaire and request for records.
• We’ll contact family within a week of receiving these to make an appointment (usually see referrals within 2-4 weeks) OR if I don’t think I can be helpful, I’ll call you to discuss other options.
  – Dr. Richard Chung sees many of our adolescent referrals.
Tips

• Collect info, then re-assess and do work-up.
• Re-shuffle your problem list 😊
  – Make your differential on the correct symptom.
• Sort symptoms/signs/previous diagnoses into known, unknown, and “thought to be known”.
• Use time-lines for complicated histories.
References


Questions?