Nine Don't-Miss Diagnoses in Young Adults

James R. Jacobs, MD, PhD, FACEP
Director – Student Health Services
The Ohio State University
Office of Student Life
Wilce Student Health Center

Don't Miss Rhabdomyolysis in Young Adults

9 Diagnoses

	Disproportionate impact on young adults	Easy to miss or misdiagnose	Immediate threat to life or organ	Sudden death in young adults
Rhabdomyolysis		•	•	
Necrotizing Fasciitis		•	•	
Hodgkin Lymphoma	•	•		
Ectopic Pregnancy			•	
WPW	•	•		•
Pulmonary Embolism		•	•	•
Peritonsillar Abscess	•		•	
Hypertrophic Cardiomyopathy	•	•		•
Testicular Torsion	•	•	•	

Don't Miss Rhabdomyolysis in Young Adults

- **Definition**
  - **Syndrome resulting from acute necrosis of skeletal muscle fibers and consequent leakage of muscle constituents into the circulation**
  - **Characterized by limb weakness, myalgia, swelling, and, commonly, gross pigmenturia without hematuria**
    - **Can include low-grade fever, nausea, vomiting, malaise, and delirium**

Don't Miss Rhabdomyolysis in Young Adults

Etiologies	Examples
Trauma	Crush injury, lightning or electrical injury, prolonged immobilization, burns
Excessive muscle activity	Strenuous exercise, status epilepticus, status asthmaticus
Increased body temperature	Heat stroke, malignant hyperthermia, neuroleptic malignant syndrome
Toxins and drugs	Ethanol, cocaine, amphetamines, PCP, LSD, carbon monoxide, benzodiazepines, barbiturates, statins, fibrates, neuroleptics, envenomation (e.g., snake, black widow, bees), quail ingestion
Infection	Many viral and bacterial infections (including influenza, Legionella, TSS); sepsis
Metabolic imbalance	Hypokalemia, hypophosphatemia, hypocalcemia, hypo- or hypernatremia
Inherited conditions	e.g., McArdle disease
Immune reactions	Polymyositis, dermatomyositis

Don't Miss Rhabdomyolysis in Young Adults

- **Keys to diagnosis**
  - **Be suspicious!**
  - **Hint: urine is dipstick positive for blood but micro negative**
  - **Serum creatine kinase > 5 times the normal limit**
    - **Also order Chem7 and serum calcium (+/- ECG)**

Don't Miss Rhabdomyolysis in Young Adults

- **Accounts for 5-15% of all cases of acute renal failure**
  - **Free circulating myoglobin**
  - **Multifactorial**
- **Electrolyte derangement can result in arrhythmias**
  - **⇧ K^+**
  - **⇩ Ca^{++}**
- **Excessive muscle swelling can result in a compartment syndrome**

Don't Miss Rhabdomyolysis in Young Adults

- **Some clinico-administrative thoughts**
  - **Maintain a high level of suspicion**
  - **Document 5P's for an affected limb(s) and thoughts about compartment syndrome**
  - **Document cardiac exam**

Don't Miss Necrotizing Fasciitis in Young Adults

Don't Miss Necrotizing Fasciitis in Young Adults

- 3 Proposed Types
  - Polymicrobial (most common)
  - Monomicrobial
  - Vibrio vulnificus (worst)

Don't Miss Necrotizing Fasciitis in Young Adults

- Rare but limb- and life-threatening soft-tissue infection
  - Characterized by rapidly spreading inflammation and subsequent necrosis of the fascial planes and surrounding tissue
- More accurately named *necrotizing soft tissue infection*
  - Mortality increases with depth of infection

Don't Miss Necrotizing Fasciitis in Young Adults

- The infection typically follows a trauma
  - Ranging from major surgery to injection sites to minor abrasion or insect bite
  - Often unnoticed
- Treatment is surgical debridement
  - Time-to-debridement is most important factor affecting mortality

Don't Miss Necrotizing Fasciitis in Young Adults

- Classic signs and symptoms
  - Blisters and bullae form and drain
    - Initially serosanguineous followed by hemorrhagic fluid
  - Skin shows violaceous discoloration before turning frankly necrotic and sloughing
    - Crepitus may be present
  - Disproportionate pain is replaced by analgesia
- Infection can spread as fast as 1 inch per hour with little change in overlying skin

Don't Miss Necrotizing Fasciitis in Young Adults

- Some clinico-administrative thoughts
  - Routinely measure or mark cellulitis boundaries
  - Tell patient and document what to expect lesion will do
  - Palpate and document beyond lesion boundaries
  - In a patient thought to have an uncomplicated cellulitis or superficial abscess who now has swelling, disproportionate pain, and evolving skin lesions, consider the possibility of necrotizing fasciitis

Don't Miss Necrotizing Fasciitis in Young Adults

- Consider the possibility
  - Tenderness beyond the margins of the visible problem
  - Pain out of proportion to the visible problem
  - Crepitus
  - Rapid worsening
- Be especially wary if this is the 2nd or 3rd visit for the same acute, initially minor skin problem

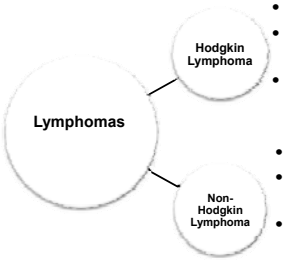
Don't Miss Hodgkin Lymphoma in Young Adults

Don't Miss Hodgkin Lymphoma in Young Adults

- Synonyms
  - Hodgkin Lymphoma
  - Hodgkin's Lymphoma
  - Hodgkin Disease
  - Hodgkin's Disease

Don't Miss Hodgkin Lymphoma in Young Adults

US Epidemiology



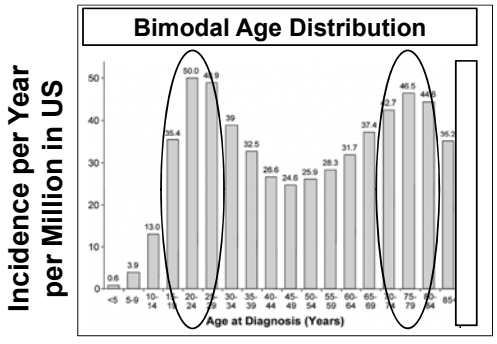
- 5 subtypes
- 8,500 cases/year
- Bimodal age distribution

- 30 subtypes
- 66,000 cases/year
- More likely in older persons

Don't Miss Hodgkin Lymphoma in Young Adults

- Definition
  - Malignant disorder of B-cells
  - Affects the reticulo-endothelial and lymphatic systems
  - Invasive disease can sometimes affect other organs and systems

Don't Miss Hodgkin Lymphoma in Young Adults



Don't Miss Hodgkin Lymphoma in Young Adults

- Presenting symptoms
  - Painless lymphadenopathy
    - Cervical and/or supraclavicular lymphadenopathy (80%)
    - Axillary and/or inguinal (somewhat less common)
  - Cough, dyspnea at rest or with exercise, or orthopnea
    - Resulting from mediastinal adenopathy
  - Fever, night sweats, or 10% weight loss/6 mo
    - So-called "B symptoms"
  - Generalized pruritis
  - Lymph node pain with alcohol consumption

Don't Miss Ectopic Pregnancy in Young Adults

Don't Miss Hodgkin Lymphoma in Young Adults

- Some clinico-administrative thoughts
  - Document node exam routinely
  - Document weight routinely
  - Persistent unexplained nodes or cough = CXR +/- node biopsy

Don't Miss Ectopic Pregnancy in Young Adults

- 2% of reported pregnancies in US are ectopic
- Ruptured ectopic is leading cause of first-trimester maternal death in developed countries

Don't Miss Ectopic Pregnancy in Young Adults

Some Risk Factors	Not Risk Factor
H/O PID	Hormonal contraception
H/O prior ectopic	Emergency contraception
H/O tubal ligation	H/O surgical abortion
H/O tubal surgery	H/O medical abortion
Age over 35	
Smoking	
IVF	
In-situ IUD	
Many lifetime partners	

Half of all cases have no identified risk factor

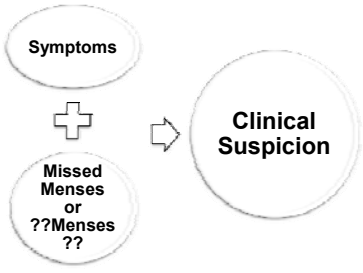
Don't Miss Ectopic Pregnancy in Young Adults

- **Presenting Complaints (examples)**
  - **Abdominal or pelvic pain**
  - **Irregular vaginal bleeding**
  - **Dizziness or weakness**
  - **Fever or flu-like symptoms**
  - **Vomiting**
  - **Syncope**
  - **Cardiac arrest**
  - **Shoulder pain**

Don't Miss Ectopic Pregnancy in Young Adults

- **Classic presentation**
  - **Pelvic or abdominal pain (99%)**
  - **Amenorrhea (74%)**
  - **Irregular vaginal bleeding (56%)**
- **Fewer than 50% of patients present with all three "classic" symptoms**
- **No statistically significant difference between presenting symptoms of unruptured ectopic vs. spontaneous abortion of IUP**

Don't Miss Ectopic Pregnancy in Young Adults



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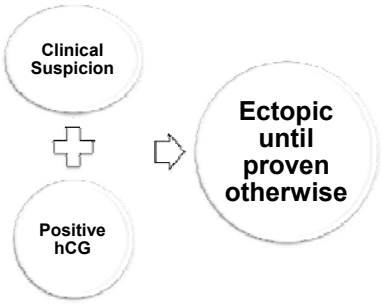


Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

Don't Miss Ectopic Pregnancy in Young Adults

- Some clinico-administrative thoughts
  - Female syncope: consider ectopic
  - IUD + positive hCG more likely to be ectopic

Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- Syndrome defined by
  - Presence of accessory cardiac conduction pathway
  - Predisposition to develop supraventricular tachydysrhythmias

Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- Bypass tracts detectable on 12-lead surface ECG
  - ≈0.20% of general population
  - 0.55% in 1st-degree relatives of patients with WPW
- No additional evidence of heart disease in ≈70% of WPW patients

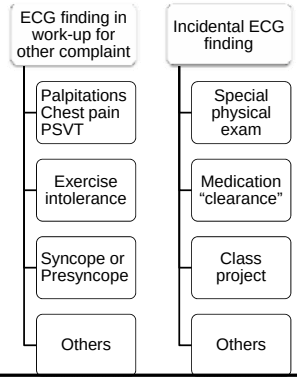
Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- Most patients present during young adulthood or middle age
- Typical: episodes of sudden-onset, sudden-offset, rapid, regular palpitations
  - Often associated with symptoms of hemodynamic compromise (e.g., dyspnea, chest discomfort, presyncope) and/or anxiety
  - Episodes persist for several seconds to several hours
- Sudden death may be the first manifestation of WPW in ≈half of patients, and it usually occurs during exercise or emotional stress

Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- High incidence of tachydysrhythmias
  - Atrial flutter (5%)
  - Atrial fibrillation (5-10%)
  - PSVT (40-80%)
- Potential to escalate to hemodynamically unstable rates or ventricular fibrillation

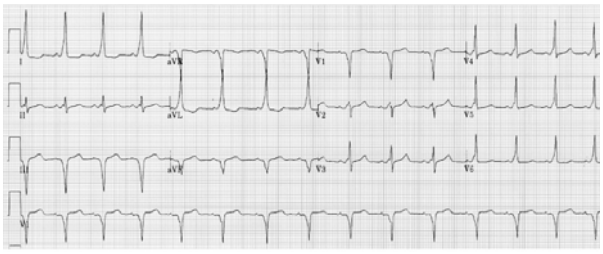
Don't Miss Wolff-Parkinson-White Syndrome in Young Adults



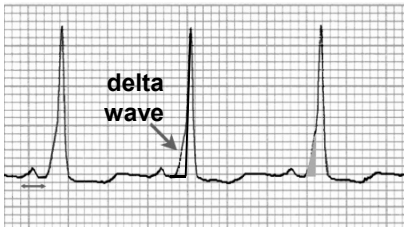
Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- "Official" ECG criteria in Adults (Surawicz et al, 2009)
  - PR interval < 120 ms during sinus rhythm
  - Slurring of initial portion of QRS that either interrupts the P wave or arises immediately after its termination
  - QRS duration > 120 ms
  - Secondary ST-TW changes

Don't Miss Wolff-Parkinson-White Syndrome in Young Adults



Don't Miss Wolff-Parkinson-White Syndrome in Young Adults



Don't Miss Wolff-Parkinson-White Syndrome in Young Adults

- Some clinico-administrative thoughts
  - Be able to recognize delta waves on 12-lead ECG
  - Intermittently symptomatic: semi-urgent referral and restrict activity
  - Incidental finding: non-urgent referral (with copy of ECG) but restrict activity
  - Calcium channel blockers, beta-blockers, digoxin, adenosine, and phenytoin are generally contra-indicated

Don't Miss Pulmonary Embolism in Young Adults

Don't Miss Pulmonary Embolism in Young Adults

- The D-dimer test is increasingly popular
  - But a negative result is [impressively] reassuring only in patients with low- to intermediate-pretest probability of clot
  - And if the D-dimer test is not available in-house with rapid turnaround, what do you do with the patient?
- How do we decide the pretest probability?
- Are there patients who don't need ANY testing?

Don't Miss Pulmonary Embolism in Young Adults

- Massive research efforts
  - Who can safely undergo no testing? Or little testing (e.g., D-dimer)
  - What is the economic and medical risk of additional testing (e.g., CT scan)?
  - What is the medical, emotional, and economic impact of false positives?
  - Is there an acceptable miss-rate?

Don't Miss Pulmonary Embolism in Young Adults

PERC Rule
Age < 50 year
Pulse < 100
Pulse ox > 94%
No unilateral leg swelling
No hemoptysis
No recent surgery
No prior DVT or PE
No oral hormone use

If clinical gestalt says maybe this is PE, but all PERC criteria are met, patient is considered so low risk that no testing is needed.

Don't Miss
Pulmonary Embolism
in Young Adults

Wells Scoring System for PE	Points
Clinical signs and symptoms of a DVT (minimum of swelling and pain on palpation of the deep veins)	3.0
Pulmonary embolism is the most likely diagnosis in the opinion of the clinician, using all available blood results, ECG, and chest xray	3.0
Heart rate >100 beats/min	1.5
Immobilization for a minimum of 3 days or surgery within last 4 weeks	1.5
Previous DVT or PE	1.5
Hemoptysis	1.0
Malignancy with treatment or palliative care in the last 6months	1.0

Wells Score	Probability of PE
<2.0	Low
2.0-6.0	Intermediate
>6.0	High

Don't Miss
Pulmonary Embolism
in Young Adults

- **Some clinico-administrative thoughts**
  - **Document scoring system**
  - **Low risk + negative D-dimer is reassuring**
  - **Rapidly evolving literature with medical and medicolegal implications**

Don't Miss
Pulmonary Embolism
in Young Adults

My assessment of where we are		
Clinical Suspicion for PE	**D-dimer**	**Dispostion**
Low	Negative	Home
Low	Positive	ER
High	Negative	ER
High	Positive	ER

Don't Miss
Peritonsillar Abscess
in Young Adults

Don't Miss Peritonsillar Abscess in Young Adults

Abscess	Usual Age	Lethal
Peritonsillar	Adolescents, adults	Death can occur from aspiration, airway obstruction, erosion into major blood vessels, or extension to the mediastinum
Retropharyngeal	< 4 yr	
Lateral pharyngeal	> 8 yr, adolescents, adults	

Don't Miss Peritonsillar Abscess in Young Adults

Common Symptoms

- Fever
- Malaise
- Severe sore throat (worse on one side)
- Dysphagia
- Otalgia (ipsilateral)

Spectrum of Exam Findings

- Erythematous, swollen soft palate with uvula deviation to contra-lateral side
- Cervical lymphadenitis and neck tenderness on affected side
- Rancid breath
- Signs of dehydration
- Ill-appearing
- Muffled voice
- Drooling
- Trismus
- Neck in slight extension

Require immediate attention to assess airway and toxicity

Don't Miss Peritonsillar Abscess in Young Adults

- **Suppurative complication of acute tonsillitis**
- **Most peritonsillar abscesses are polymicrobial**
  - **And thus can present even after several days of appropriate antibiotic treatment for "strep throat"**

Don't Miss Peritonsillar Abscess in Young Adults

- **Tonsillectomy**
  - **Single episode of PTA with successful intervention/resolution typically does not meet criteria for interval tonsillectomy**
    - **Recurrent episodes usually do**
  - **History of tonsillectomy does NOT R/O possibility of subsequent peritonsillar abscess**

Don't Miss
Peritonsillar Abscess in Young Adults

- Some clinico-administrative thoughts
  - Sore throat + drooling + voice change = emergent evaluation
  - Already "on antibiotics" ≠ low priority for re-visit
  - For EVERY sore throat document presence or absence of PTA findings
  - No such thing as a simple sore throat

Don't Miss
Hypertrophic Cardiomyopathy in Young Adults

- Inappropriate myocardial hypertrophy
  - Often symmetric
  - Occurs in the absence of an obvious inciting stimulus
- Genetic disorder
  - Autosomal dominant
  - Variable penetrance and variable expressivity

Don't Miss
Hypertrophic Cardiomyopathy in Young Adults

Don't Miss
Hypertrophic Cardiomyopathy in Young Adults

- Sudden death can be the first manifestation
  - Ventricular fibrillation or unstable ventricular tachycardia
  - Leading cause of sudden death in young people
  - Best known as the cause of sudden death in high school and college athletes

Don't Miss Hypertrophic Cardiomyopathy in Young Adults

- Harmon et al: Incidence of sudden cardiac death in NCAA athletes. *Circulation* 2011
  - 187 nonmedical or trauma
  - 45 of 80 medical deaths were cardiac
    - B-ball > F-ball > Swim > Lacrosse/CC
    - Basketball
      - Black male 1:4,000; White male 1:13,000; Female 1:38,000
      - Division I male 1:3,000

Don't Miss Hypertrophic Cardiomyopathy in Young Adults

- Most patients with HCM have a normal physical exam
  - IF a murmur is heard it is typically only when patient stands or performs Valsalva
    - Late-systolic ejection murmur best heard at the left sternal border radiating to the aortic and mitral areas but not into the neck

Don't Miss Hypertrophic Cardiomyopathy in Young Adults

- Major risk factors for sudden death in HCM include
  - Prior cardiac arrest
  - Unexplained syncope
  - Family history of premature sudden death
  - Nonsustained or sustained VT
  - LV wall thickness ≥ 30 mm
  - Abnormal blood pressure response to exercise

Don't Miss Hypertrophic Cardiomyopathy in Young Adults

Screening for HCM: 9 questions we need to ask (Published widely, including deWeber, 2009)
Have you ever passed out or nearly passed out during exercise?
Have you ever passed out or nearly passed out after exercise?
Have you ever had discomfort, pain, or pressure in your chest during exercise?
Does your heart rate race or skip beats during exercise?
Has your doctor ever told you that you have a heart murmur?
Has a doctor ever ordered a test for your heart?
Has anyone in your family died for no apparent reason?
Does anyone in your family have a heart problem?
Has any family member or relative died of heart problems or of sudden death before age 50?

Clinician's response to affirmative answers remains largely a matter of professional judgment

Don't Miss
Hypertrophic Cardiomyopathy
in Young Adults

- Active research and controversy about whether to routinely include 12-lead ECG in preparticipation screening of young athletes
- And then there's preparticipation echocardiograms…

Don't Miss
Hypertrophic Cardiomyopathy
in Young Adults

- Some clinico-administrative thoughts
  - Auscultation for physical exam should be supine and upright (or Valsalva)
  - Follow guidelines for high school and college athletic physicals
  - Syncope during or after exercise is ominous

Don't Miss
Hypertrophic Cardiomyopathy
in Young Adults

My Current Interpretation of the State-of-the-Art			
Cardiac Exam	**History**	**ECG**	**Disposition**
Normal	Normal	No	No restrictions
Abnormal and Unexplained	Regardless	+/-	Refer and restrict
Normal	Nuisance answers	Yes-Normal	No restrictions
Normal	Nuisance answers	Yes-abnormal	Refer +/- restrict
Normal	Concerning	+/-	Refer and restrict

Don't Miss
Testicular Torsion
in Young Adults

Don't Miss Testicular Torsion in Young Adults

- Terminology
  - "testicular" torsion is actually torsion of the spermatic cord
- Types
  - Intravaginal (most common)
    - Classic "bell-clapper" abnormality
  - Extravaginal
  - Mesorchial (very rare)

Cannot be distinguished clinically, so we discuss as a single entity

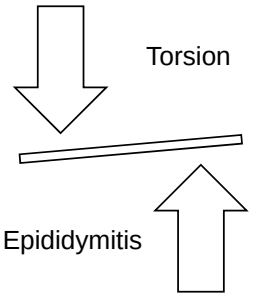
Don't Miss Testicular Torsion in Young Adults

- 1:4,000 males < 25 yo
  - ≈ 60% cases < 21 yo
    - Oldest reported case 69 yo
  - Bimodal distribution with two peaks
    - Neonatal period
    - ≈ age 13
- Bilateral in up to 2% of cases

Don't Miss Testicular Torsion in Young Adults

- Most cases occur in the absence of any precipitating event
  - 5-8% of cases result from trauma
  - Other risk factors include
    - onset of testicular growth in puberty
    - h/o cryptorchidism
    - testicle with a horizontal lie at baseline
    - spermatic cord with long intrascrotal portion

Don't Miss Testicular Torsion in Young Adults



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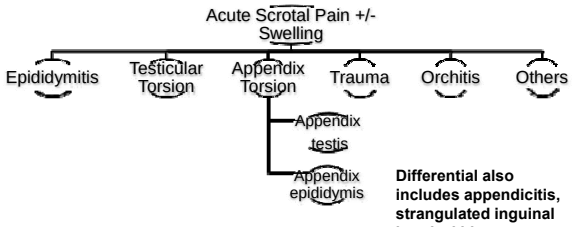

Differential also includes appendicitis, strangulated inguinal hernia, kidney stone, and others.

Don't Miss Testicular Torsion in Young Adults

The acute severe presentation is relatively straightforward…ER, even if it's just for pain control

Don't Miss Testicular Torsion in Young Adults

Classic Signs , Symptoms, and Findings			
	Epididymitis	**Testicular Torsion**	**Appendix Torsion**
Pain	Gradual, focal	Sudden, diffuse	Gradual, focal
Urinary Sx	Maybe	Absent	Absent
Cremasteric	Intact	Absent	Intact
N/V	Unusually	Common	Rare
Lie	Vertical	Horizontal	Vertical
Prehn's sign	Positive	Negative	??

Sensitivity and specificity of these are so poor as to be essentially worthless

Don't Miss Testicular Torsion in Young Adults

- **What about subacute?**
  - **Onset more than a few hours out, or maybe pain is subsiding**
- **What about non-acute?**
  - ***I've had this pain for about a week now.***
  - ***Something like this happened for a few minutes two other times.***

Don't Miss Testicular Torsion in Young Adults

- If, in spite of your best history and physical exam, it is not clear
  - Don't just send to ER or imaging center (for Doppler U/S or radionuclide imaging)
    - If he is in severe pain you should have sent him already as an acute scrotum
    - If he is not in severe pain he will languish
  - Consult with urologist and/or radiologist

Don't Miss Testicular Torsion in Young Adults

- Some clinico-administrative thoughts
  - No physical exam finding can rule in or rule out torsion
  - Always re-think a diagnosis of epididymitis
  - History of intermittent severe scrotal pain deserves referral

Don't Miss Testicular Torsion in Young Adults

- Intermittent torsion occurs
  - Can last minutes or hours
  - Partial torsion also occurs and can damage testicle
- Even if patient is pain free in the office, ask about prior episodes
  - If there is history suggestive of intermittent torsion, refer
  - Consult urologist prior to ordering an asymptomatic imaging study

More Don't-Miss Diagnoses

- Lemierre syndrome
- Testicular cancer
- Tick-borne illness
- Marfan syndrome
- Pneumothorax
- Aortic stenosis
- Carbon monoxide toxicity
- Contact lens injury
- Tropical diseases
- TB
- HIV
- Syphilis
- APAP toxicity
- Bacterial meningitis
- Appendicitis
- PID
- And many more…