High Yield Psychiatry

Shelf Exam Review

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A patient is brought in by his identical twin brother stating he has been sleeping little for the past 8 days, had sex with 15 different women, and talked in a pressured manner about maxing out his credit cards "starting a business that couldn't fail".

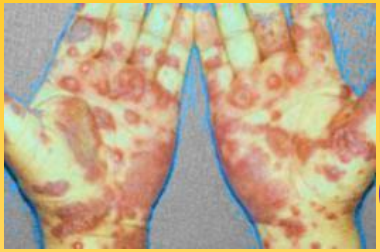
- Diagnosis? Manic Episode → bipolar I if cycled w/ depressive episodes
- Incidence in the population? ~1%
- Risk for same Dx in brother? 80-90%
- If these sxs occurred for the 1st time in a 75 y/o patient? Look for a medical cause. *Right frontal hemisphere stroke*
- Medications to AVOID? SSRIs and TCAs (can trigger mania)
- Medications to start in this patient? Haloperidol or clonazepam for acute agitation or delusions.
  Lithium, valproic acid or carbamazepine for maintenance.

- Patient taking Advil develops n/v/d, coarse tremor, ataxia, confusion, slurred speech.

Lithium Toxicity
Precip by NSAIDs.
Better pain meds are aspirin or sulindac.

- Possible EKG findings? T-wave flattening or inversion + U waves
- Tx? Fluid resuscitation. Emergent dialysis if >4 or kidney dz
- Major Side Effects? Weight gain and acne, GI irritation, cramps
- MOA? Suppresses inosital triphosphate
- Therapeutic levels? 0.6-1.2
- Medical monitoring? Li level q4-8wks, TFTs q6mo, Cr, UA, CBC, EKG
- Contraindications for use? Severe Renal Dz, MI, diuretics or digoxin, MG, pregnancy or breastfeeding.
- Problems in preggos? Ebstein's anomaly = malformed tricusp, atrializes part of RV. If taken during 1st tri

- Preferred treatment for bipolar in preggos? Clonazepam. Esp 1st trimester
- Bipolar + elevated LFTs and hepatitis? Valproate. Also can cause n/v/d, skin rash
- Bipolar + Steven's Johnson Syndrome?



Lamotrigine
(less likely carbamazepine

- Bipolar + agranulocytosis? Carbamazepine. Check CBC regularly
  - If ANC <2000? Monitor closely w/ weekly CBC
  - If ANC <1000? D/C the med
- Bipolar + ↑AFP in a 20wk preggo? Could be Valproate or Carbamazepine → NTD. Repro-age F should take 4g daily
- Most common complication of carbamezapine? Rash.
- Therapeutic levels of valproate? 6-12
- Therapeutic levels of carbamezapine? 60-120

A woman comes in complaining of decreased appetite and 5lb weight loss, no longer enjoys knitting, insomnia and decreased energy, unable to concentrate and feeling guilty for 2 weeks.

- Most important 1st question? Assess for suicidal ideation.
- RF for this? *Prior attempt*, >45, white, male, serious illness, detailed plan, no support, lack of support, ETOH/drugs
- Seen on polysomnogram? Shortened REM latency, more freq REM
- Atypical lab test? Dexamethasone suppression test → failure to suppress
- Medications that might cause this? IFN, beta-blockers, αmethyldopa, L-dopa, OCPs, ETOH, cocaine /amph withdrawal, opiates.
- Medical diseases that might cause this? HIV, Lyme, Hypothyroidism, Porphyria, Uremia, Cushings Dz, Liver disease, Huntington's, MS, Lupus, L-MCA stroke

- Patient who is eating more, gaining weight, sleeping more and has *leaden paralysis* in the morning.
- 1 month after death of her child, a mother feels guilty, can't sleep, concentrate, eat, or enjoy her interests.
- 4 months after the death of her chihuahua, a woman still feels guilty, can't sleep, concentrate, eat, or enjoy her interests.

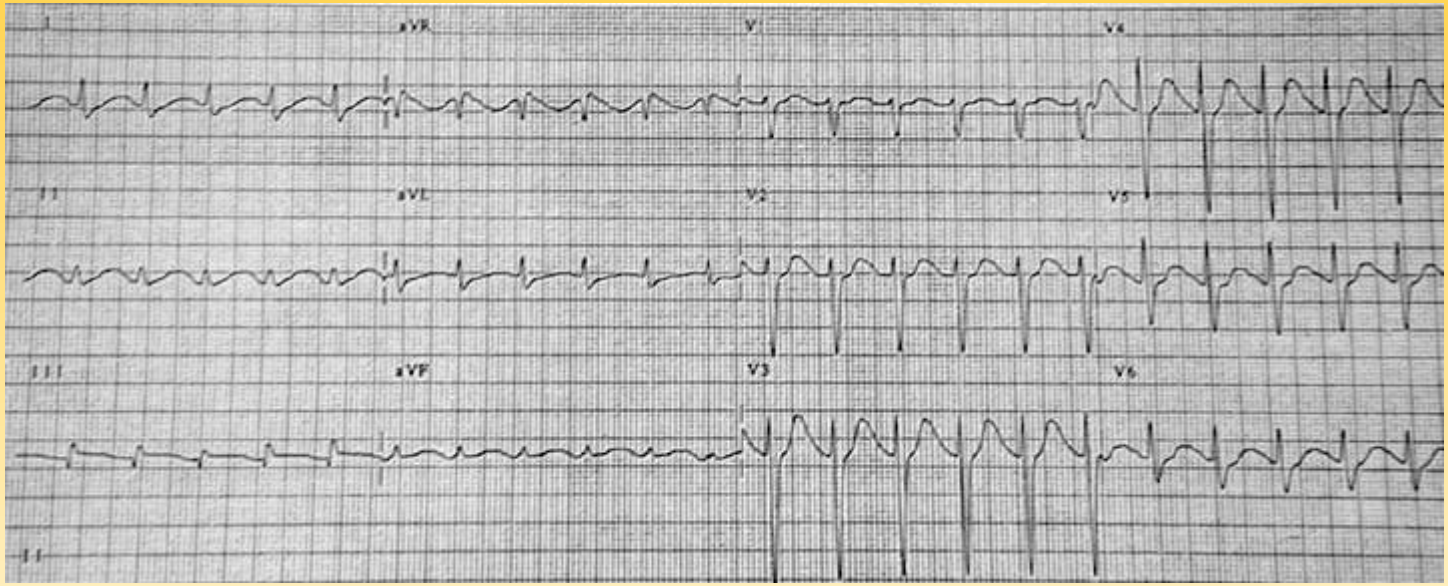
Atypical Depression.
Are hypersensitive to rejection, can affect social fxning.
*Best treated w/ MAOIs.

Uncomplicated Bereavement.
V-code on DSM-IV
No suicidal ideation (other than thoughts of wanting to be w/ loved one). No psychosis (other than hearing/seeing loved one)
*Rarely tx w/ antidepressants for sxs

Adjustment Disorder.
Sxs w/in 3mo of stressor out of proportion. Can't persist longer than 6mo.
*Best treated w/ psychotherapy.

- 1st line for MDD- **SSRIs**. Also indicated for OCD, bulemia, anxiety, PTSD or premature ejaculation
  - Has most drug-drug interactions Paroxetine
  - Don't have to taper when stopping Fluoxetine
  - Fewest drug-drug interactions Citalopram
  - HA, n/v/d, dizziness and fatigue when stopping suddenly. **5HT discontinuation syndrome**. Most common w/ sertraline and fluvoxamine
- Myoclonic jerks, tachycardia, high BP, hyperreflexia, n/v/d. **5HT syndrome**. If SSRI + MAOI
- What if loss of erection, ejaculation? Switch to buproprione (DA/N-RI)
  - Contraindications to use? Bulemia, alcoholics, epileptics
- Erection lasting >3 hours? Likely caused by trazodone.
- Good for old, skinny, sad ladies? Mirtazepine. ↑appetite and sleep
- Avoid in hypertensive patients? Venlafaxine (SNRI). Don't take w/ St. Johns Wart
- Pounding head, flushing, nausea, myoclonus after eating cheese, drinking red wine, taking decongestant or merperidine? Hypertensive crisis w/ MAOI. Tx w/ 5mg IV phentolamine

A kid ate some unidentified pills out of grandma's purse. Grandma has HTN, HLP, fibromyalgia, insomnia and peptic ulcer disease. He now has dry mouth, tachycardia, vomiting, urinary retention, and seizures.



"Widened QRS complexes and prolonged QT interval"

- What did the kid ingest? Tricyclic Antidepressant
- Most common cause of death? Arrhythmia → torsades, v-fib, death
- Treatment? Activated charcoal if ingestion w/in 1-2hrs. Give IV sodium bicarbonate. (helps met acidosis and cardioprotective)

A smelly 20 y/o college kid's grades have been declining over the past 2 semesters as he keeps to himself, has flattened affect and no motivation. For the past 6wks, he has locked himself in his dorm room stating President Obama "put a hit on him". He was told this by 2 voices having a discussion in his head.

- Diagnosis? Schizophrenia, Paranoid type (MC and best prog)
- Prevalence? 0.5-1%
- Risk for MZ twin? 50%
- Sibling? 10%
- Neurobiology? Positive Sxs-excess DA in limbic area binding D2 recept.
  Neg Sxs- decr DA in prefrontal cortex/meso-cortical tract
  *This is why typical antipsychotics make negative sxs worse.

- A patient has delusions, hallucinations, and flattened affect for 3 weeks.
  - For 3 months? Schizophreniform Disorder (>1m, <6mo)

Brief Psychotic Disorder (>1wk, <1mo)

- A patient has had persecutory delusions for the past 3 years. 6 months ago he started having sadness, guilt, insomnia, ↓concentration, SI.

Schizoaffective Disorder. (delusions/hallucinations for >2wks in absence of mood ss)
*Tx w/ Atypical antipsychotics + SSRI if depression and + Li if manic.

- A patient has had MDD for 3 years and reports hearing voices telling him he is worthless and to kill himself.

MDD with Psychotic Features. Delusions are typically mood congruent.
* Tx w/ Atypical antipsychotic + SSRI or ECT (esp in preggos)

- A man is convinced Miley Cyrus is in love with him but is otherwise functional.

Delusional Disorder. Erotomanic type. Non-bizzare.
Tx w/ therapeutic relationship + meds

- DOC for acute agitation or psychosis? IM haloperidol.
- MOA? D2 receptor antagonist. @ mesolimbic tract → helps + sxs. Causes hyperprolactinemia and EPS.
- Low Potency? Chlorpromazine and Thioridazine. Less EPS more anti-Ach
- High Potency? Haloperidol and Fluphenazine. More EPS.
- If patient has a history of medication non-adherence? Can give decanoate forms ever 2-4wks.
- Purple grey metallic rash over sun-exposed areas and jaundice? Chlorpromazine
- Prolonged QTc and pigmentary retinopathy? Thioridazine

- Pt wakes up with eyes "stuck" looking up or head "stuck" turned to the side.

Acute Dystonia. (<12hrs).
Tx w/ benztropine or diphenhydramine

- Pt reports feeling like they "always have to move".

Akathesia. (30-90 days).
Tx w/ propranolol (1st line) or benzo

- Coarse resting tremor, masked facies, unsteady gait, bradykinesia.

Parkinsonism. (>6mo)
Tx w/ benztropine/diphenhydramine, amantidine or bromocriptine. NOT L-dopa!!

- After 10 years on fluphenazine, tongue movments and gimacing.

Tardive Dyskinesia. (>years)
Tx by stopping antipsychotic and switching to and atypical or clozapine.

- W/in hours of a haloperidol injections, pt has ↑CPK, T = 103F, rigidity, autonomic instability, and delirium.

Neuroleptic Malignant Syndrome.
1st- d/c the offending med.
2nd- cooling blankets and dantroline Na or bromocriptine (2nd line).
Remember that metoclopramide, compazine and droperidol can cause.

- Atypical agent w/ highest risk for EPS and ↑prolactin? Risperidone. But comes in depo shot
- Weight neutral but prolongs the QTc? Ziprazodone.
- Weight neutral but increases akathesia? Aripiprazole.
- Most assoc w/ weight gain? (but #1 S/E is sedation.) Olazepine
- Causes orthostasis and cataracts? Quetiapine (alpha blocking properties)
- Good for tx-refractory schizophrenia? Clozapine
  - Most Common S/E- Sedation, weight gain, ↑blood sugar and lipids
  - Most Dangerous S/Es- Agranulocytosis, decreased seizure threshold.
  - Monitoring? CBC → ANC qweek for 6mo and x2wks for next 6mo. D/c if WBCs<3000 or ANC<1500

A 28 y/o female is brought in by EMS complaining of shortness of breath, palpitations and chest pain. She smokes 1 PPD and her only medication is OCPs. She had one of these attacks previously while grocery shopping. She shares with you that she is so afraid of having another one she rarely leaves her house.

- What is your next step?

  EKG, cardiac enzymes, echocardiogram, TSH or T4, urine drug screen,

- Drug regimen of choice?

  Alprazolam or clonazepam low dose PRN short term, but SSRIs are the preferred drug

  *Don't give benzos to drug addicts, COPDers, or restrictive lung disease.

- She is brought in 3mo later with sxs of a temp of 101, convulsions, confusion and hypertension. She recently lost her perscription drug coverage.

  Acute benzo withdrawal reaction.
  Similar to DTs.
  Tx w/ diazepam or chlordiazepoxide + haloperidol if psychotic.

- MS4 w/ deathly fear of flying that inhibits her from interviewing at the program of her dreams.
- MS3 w/ deathly fear of presenting a case in grand rounds b/c she is afraid the surgeons will laugh at her.
- MS2 keeps to herself and doesn't talk with peers b/c she is afraid they will laugh at her.
- MS1 is having difficulty falling asleep b/c she keeps thinking about failing biochem. In class she cannot concentrate b/c she worries her boyfriend will leave her. Sxs lasting >6mo

Specific Phobia.
Best Tx is CBT w/ flooding or exposure/extinction.
Can give benzos for situational use.

Social Phobia.
Best Tx is propranolol to stop hyperarousal and benzo.

Avoidant Personality Disorder.
Best Tx is CBT

Generalized Anxiety Disorder.
Best Tx is Buspirone (5HT 1a partial agonist), but must give benzos to bridge b/c it takes >3wks to work.

18y/o who just started college has declining grades. He states he can't make it to class on time because he spends 2-3 hours scrubbing in the shower each morning. He knows this is excessive but on days he takes shorter showers, he states he can "feel the bacteria" and worries about contracting an illness.

- Dx? Obsessive Compulsive Disorder
- Comorbid Condition? High prevelance of vocal-motor ticks and 5-7% of OCD pts have full blown Tourettes.
- Tx? Clomipramine is gold standard
  SSRIs are first line.

A 25 y/o sexual assault survivor comes to you with a 6wk history of recurrent nightmares of when she was raped at knifepoint. She now avoids situations where unknown men will be present, to the point that she had to quit her job at a bank. She reports being "jumpy" anytime she hears footsteps behind her.

- Dx? Post Traumatic Stress Disorder
- Tx? Sertraline or paroxetine. Combined w/ CBT. Prazosin for NMs
- If same sxs, but only present for 3wks? Acute Stress Reaction
- If same sxs, but in response to a bad breakup? Adjustment Disorder

- A 54 y/o RN presents w/ a history of 2mo of diarrhea and abd pain. He has presented to 4 other hospitals w/ the same complaint. Colonoscopy reveals pigmentation in the wall of the colon

Munchausen Syndrome.
More severe than simple factitious d/o b/c they actually induce sxs. (in this case, w/ laxative abuse).
They do it for *primary gain*.

- A concerned mother presents with her 15mo baby who is having recurrent seizures. She requests an MRI, sleep deprived EEG with intracranial leads.

Munchausen Syndrome by proxy.
A form of child abuse!
10% of children die before reaching adulthood.

- A 45 y/o unemployed man is involved in a car accident. He sues the driver stating he has nerve damage to his legs that keeps him from walking. Video evidence shows him dancing at a club the night before.

Malingering.
Goes as a V-code
Associated w/ antisocial personality disorder
They do it for *secondary gain*.

A 18 y/o F presents with no menstrual cycle for 3mo. A pregnancy test is negative but her BMI is calculated to be 17. Her teeth are eroded and she has calluses on her knuckles (Russel sign).

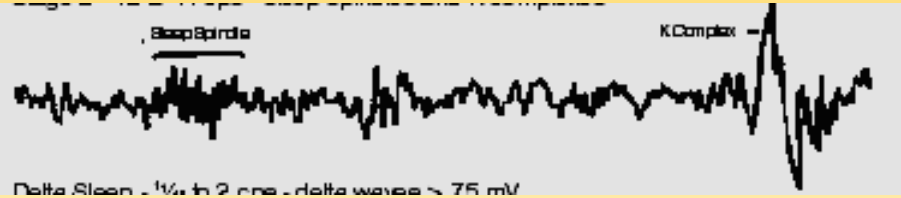
- Laboratory abnormalities-
  - Vital signs Hypotension, Bradycardia, Hypothermia
  - CBC Leukopenia
  - Chemistry High HCO3, low Cl, low K, high carotene, high LFTs and amylase
  - TFTs normal
  - Fasting Lipid Profile High cholesterol
  - Hormones High cortisol, low LH/FSH, low estrogen
- Long term complications- Osteoporosis
- Most common cause of death- Heart disease. Then suicide.
- Treatment- Admit them to maximize nutrition. SSRI's help bulemia, anoexia needs intensive counseling.
- Complications of Treatment-

Re-feeding syndrome = low PO4, low Mg, low Ca and fluid retention.

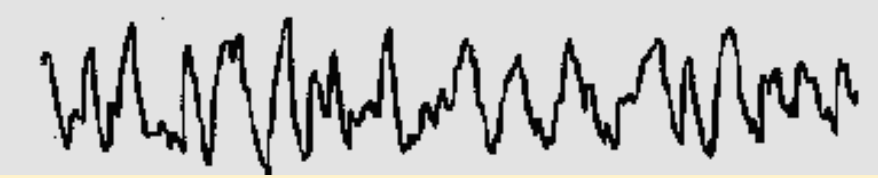
Sleep EEGs

Awake

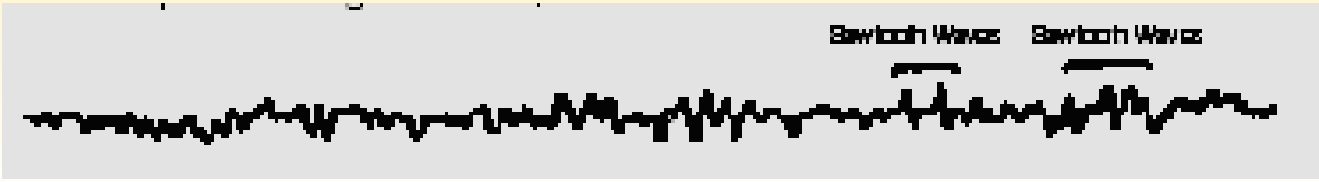
Stage 1



Stage 2



Slow wave sleep-
Stage 3 <50% delta, Stage 4 >50%
Sleep walking/talking/night terrors



REM.
Skeletal muscle paralysis

- Trouble falling asleep or staying asleep causes impairment in fxn >1mo.
- As falling asleep, feel creepy-crawlies on legs, better when they get up and move.
- Daytime sleepiness and depression in a big fat guy with a big neck.
- Irresistible attacks of refreshing (REM) sleep. Upon intense emotion, they lose muscle tone or have hallucinations as waking of falling asleep.

Insomnia.
Educate about sleep heigyne 1st, then try benzos (reduce sleep latency and incr SWS and REM). Zolpidem, zaleplon, escopiclone are GABAa recp

Dyssomnia NOS.
R/o medical causes 1st → Fe-def anemia or chronic kidney dz. Neuropathy.
Tx w/ ropinirole or pramipexole (Da-ag)

Obstructive Sleep Apnea.
Goes on axis III, "breathing related sleep d/o" goes on axis I.
Need polysomnogram to diagnose → >10 hypopneic/apneas per hour. Need CPAP to reduce pulmonary HTN.

Narcolepsy.
Tx w/ scheduled naps and Modafinil.

- 30 y/o man and is wife present for couples counseling. He constantly accuses her of cheating. He's in a feud w/ the neighbor b/c he feels they are attacking his character when they say they like his flowerbeds.
- 30 y/o man, never been married or have any close friends. Works as a night security guard and in his free time works on his model ships in his basement.
- 30 y/o man, never been married or have any close friends because "people make him uncomfortable". He is unemployed because he spends his time reading books on how to communicate with animals so he can "be at one with nature".

Paranoid PD
Low dose anti-psychotics can help paranoid behavior.

Schizoid PD
Distinguish from Avoidant b/c they don't WANT relationships

Schizotypal PD
Distinguish from Schizoid by magical thinking/ interests.
Distinguish from Schizophrenia by lack of delus/hallu

- 25y/o man comes to court mandated counseling for beating his girlfriend. He was kicked out of high school for fighting & just got out jail for stealing a car.
- His girlfriend has a hx of unstable relationships, has superficial cuts on both wrists, is impulsive in her spending and sexual practices.
- 26 y/o MS2 is asked by Nan Clare to seek counseling. Her classmates complain that she dresses too provocatively to class. She recently tried to seduce a professor.
- A 22 y/o MS1 doesn't feel like he needs to come to any classes or labs because he "already has the brilliance to be a doctor. He refuses to talk to Nan Clare about this, instead insisting to deal directly with President Henrich.

Antisocial PD. 2/3 have substance abuse.

Borderline PD. Commonly use splitting.

Histrionic PD. Look for substance abue or eating d/o

Narcissistic PD. Can be confused w/ hypomania b/c of grandiosity. Give individual thearpy

- 30 y/o woman has no friends and avoids happy hours with her coworkers b/c she fears ridicule and rejection. She feels “no one would want to be friends with me”.
- 30 y/o woman has jumped from one relationship to another because she “doesn’t do well alone”. She calls her friends and family >20x a day to get their input on her daily decisions.
- 25 y/o MS4 spends more time color coding her notes and textbook highlighting than actually studying. She makes lists and study schedules 3 times per day. People don’t like to work with her because she is so “anal”.

Avoidant PD. Can tx social phobia sxs w/ b-blocker or SSRI

Dependent PD. Look for co-morbid depression and anxiety. SSRI

Obsessive Compulsive PD. Different from OCD b/c the actions are “ego-syntonic”

78 y/o lady is brought in from her nursing home for altered mental status. She sleeps more during the day and becomes agitated at night- reporting seeing green men in the corner. She also complains of pain upon urination.

- First step? In her case, UA and culture.
  Work up also includes glc, Na, blood cultures, B12, RPR
  Make sure to look at med list- benadryl, opiates, Bzs
- Biggest risk factor? Age. Underlying dementia is the 2nd biggest
- Other common causes? Acute substance withdrawal. Look for it on the 2nd or 3rd post-op day in alcoholic.

- EEG findings? Diffuse background slowing of background rhythm. Psychosis has normal EEG.
- Treatment? Reduce excessive stimuli, calendar and clock to orient patient. Stop unnecessary meds. Give haloperidol if agitated.

A 78 y/o female presents with memory loss…

- Aphasia, apraxia, gets lost while driving? **Alzheimer's Dementia**. MC type. On MMSE, prompting does not ↑recall
  - Pathology? Global brain atrophy. B-amyloid plaques or tau tangles
  - Genes? APP (on chr 21), ApoE E2
  - Treatment? Donepezil, rivastigmine, galantamine (diarrhea). Memantine
- Becomes more sexually explicit, apathy. **Frontotemporal Dementia**. (Pick's Dz).
  - Pathology? Lobar atrophy. Intra-neuronal silver staining inclusions.
  - Treatment? Olanzepine for severe disinhibition.
- Fluctuation in consciousness, visual hallucinations and shuffling gait. **Lewy Body Dementia**
  - Pathology? Intra cytoplasmic Alpha-synuclein inclusions in neocortex
  - Treatment? Give Ach-Ease inhibitors. NOT L-dopa. Avoid neuroleptics.

A 78 y/o female presents with memory loss…

- Sudden, step-wise decrease in memory/cognitions. **Vascular Dementia**.
- Loss of vibration sense, labile affect. Pupil that accommodates but doesn't react. **Tertiary Syphilis**.
  - Dx? +RPR, VDRL. Do spinal tap to look for spirochetes.
  - Tx? IV penicillin. If Pen-allergic, must desensitize.
- Myoclonus, startle response, seizures. Recently had a corneal transplant. **Creutzfeldt Jakob**.
  - Pathology? Spongiform encephalopathy
  - EEG findings? *Triphasic bursts*
- Incontinence, gait disturbance/freq falls, and rapidly developing dementia. **Normal Pressure Hydrocephalus**.
  - Dx? CT/MRI shows hydrocephalus, spinal tap shows nl opening pressure
  - Tx? Ventriculoperitoneal shunt improves cognitive fxn in 50-67% of pts

A 50 y/o known alcoholic presents to the ER with tonic clonic seizures. BP 180/110, HR 118, T 100.1.

- How long since his last drink? ~12-24hrs. (bimodal peak at 8 and 48hrs)
- How long till he develops confusion, fluctuations in consciousness and the feeling of ants crawling on him? ~48-72hrs since last drink is when delirium tremens usually start.
- His blood alcohol level is 225mg/mL. How long till its out of his system? ~9hrs, Alcohol is metabolized by zero order kinetics (same amt/unit time = 25mg/hr)
- If his medications included propranolol, lactulose, and allopurinol, what would be the best sign to monitor for his withdrawals? Beta-blockers mask the signs of autonomic hyperactivity, but you can follow hyperreflexia to dose the benzos during w/drawal.

- Best initial treatment of our patient?

Diazepam or chlordiazepoxide b/c they have 80 & 120hr ½-lives respectively.

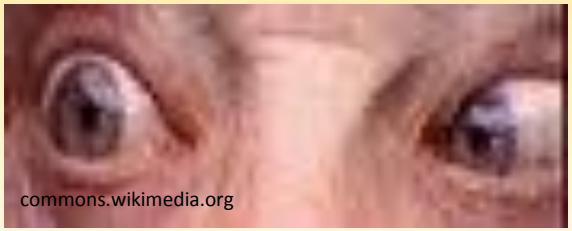
- What if he's a Child's class C cirrhotic?

Lorazepam, oxazepam or temazepam b/c they are glucuronidated prior to elim

- Most specific test for ETOH consumption in the past 10 days?

Carbohydrate-deficient transferrin. Less specific- elevated GGT and AST more than twice ALT.

- Our next patient comes in w/ confusion, ataxia, and you find this on physical exam: Dx?



Wernicke Encephalopathy.
Caused by thiamine deficiency
Give thiamine 1st, then glucose containing fluids.

Can progress to Korsakoff's syndrome (irreversible damage to mamillary bodies, etc)- apathy, anter/retrograde amnesia and confabulation. Can see MB atrophy on MRI

  - Best 1st step?

A patient is brought into the ER in a non-responsive state. His BP is 100/60, HR is 50, RR is 6. He has multiple track marks on his arms.

- Best first step?

  Intubate the patient. Then give IV or IM naloxone (full mu-opiate antagonist)

- You realize his pupils are dilated. Does that change your dx?

  No. The hypoxia 2/2 respiratory depression can cause hypoxia

- What sxs to you expect as he starts to withdraw?

  Joint and muscle pain, photophobia, goosebumps, diarrhea, tachycardia, HTN, GI cramps, dilated pupils, anxiety/depression

- Treatment?

  Clonidine for autonomic sxs, ibuprofen for muscle cramps, loperimide for diarrhea.
  Methadone, buprenorphrine or Naltrexone can be used for long-term dependence.

- Pt presents with horizontal nystagmus, dilated pupils, ataxia and acute psychosis.

  Hallucinogen (PCP) intoxication. Can use haloperidol for acute psychosis.

- Pt presents s/p MVC with injected conjunctiva, sedation and is asking for Doritos (cool ranch plz).

  Cannabis intoxication.

- Pt presents with SI, hypersomnia, depression and anergia.

  Cocaine/Amphetamine withdrawal.

- Pt presents with dilated pupils, seizure, tachycardia and HTN.

  Cocaine/Amphetamine intoxication

  - Best 1st test? EKG then urine tox screen. Tx seizure w/ lorazepam
  - Tx of HTN and tachycardia? Calcium channel blocker. Beta-blockers are CONTRAINDICATED!

Childhood Development

	Erikson	Piaget	Freud
Birth – 1 year	Trust vs Mistrust	**Sensorimotor**- control motor function- object permanence	Oral
1 year-3 years	Autonomy vs Shame		Anal
3 years-5 years	Initiative vs Guilt	**Preoperational**- Egocentric	Phallic
6 years- 11 years	Industry vs Inferiority	**Concrete Operational**- Death is permanent	Latent
11 years- adolescence	Identity vs role diffusion	**Formal Operational**- Think abstractly, deductive reasoning, hypothetical thinking.	Genital
21 years – 40 years	Intimacy vs isolation		
40 years- 65 years.	Generativity vs stagnation		
> 65 years	Integrity vs Dispair		

An 11 year old boy is evaluated for developmental delay, poor school and social performance. Formal IQ testing reveal his IQ to be 50. He has a macrocephaly, long face and macroorchidism

Mild- 55-70
Moderate- 40-55
Severe- 25-40
Profound- <25

- What degree of mental retardation?

Moderate.

- What is average and standard deviation for IQ?

Average is 100, Standard deviation is 15

- Where does it go in the DSM-IV?

Axis II

- What is the most likely cause in this case?

Fragile X
X-linked dominal inheritance
CGG repeats w/ anticipation
Cx = Seizures, MVP, dilation of the aorta, tremors, ataxia, ADHD-like behavior.
MC cause of inherited MR.

A newborn baby has decreased tone, oblique palpebral fissures, a simian crease, big tongue, white spots on his iris

Down's Syndrome

- What can you tell his mother about his expected IQ?

  He will likely have mild-moderate MR. Speech, gross and fine motor skill delay

- Common medical complications?
  - Heart? VSD, endocardial cushion defects
  - GI? Hirschsprung's, intestinal atresia, imperforate anus, annular pancreas
  - Endocrine? Hypothyroidism
  - Msk? Atlanto-axial instability
  - Neuro? Incr risk of Alzheimer's by 30-35. (APP is on Chr21)
  - Cancer? 10x increased risk of ALL

- Café-au-lait spots, seizures large head. Autosomal dominant — Neurofibromatosis
- Coarse facies, short stature, cloudy cornea. Autosomal recessive. — Hurler Syndrome
- Broad, square face, short stature, self-injurious behavior. Deletion on Chr17 — Smith Magenis
- Hypotonia, hypogonadism, hyperphagia, skin picking, agression. Deletion on paternal Chr15. — Prader-Willi
- Seizures, strabismus, sociable w/ episodic laughter. Deletion on maternal Chr15. — Angelman
- Elfin-appearance, friendly, increased empathy and verbal reasoning ability. Deletion on Chr7. — Williams

- ADHD-like sxs, microcephaly, smooth philtrum. Most common cause of mental retardation.

Fetal Alcohol Syndrome

- Seizures, chorioretinitis, hearing impairments, periventricular calcifications, petechiae @ birth, hepatitis.

Congenital CMV infection.

- Seizures, hearing impairments, cloudy cornea/retinitis, heart defects, low birth weight.

Congenital Rubella Syndrome

- Abnormal muscle tone, unsteady gait, seizures, mental retardation or learning disability.

Cerebral Palsy from birth asphyxia.

- IUGR, hypertonia, distinctive facies, limb malformation, self-injurious behavior, hyperactive.

Cornelia de Lange

- Coloboma, heart defects, choanal atresia, growth retardation, GU anomalies, ear deformity and deafness. Chr 8.

CHARGE

- Autism spectrum sxs, heart disease, palate defects, hypopastic thymus, hypoCa. Chr 22 deletion.

DiGeorge

- Vomiting, seizures, lethargy, coma. Acidosis w/ stress, illness. Causes neurological damage.

Maple Syrup Urine Disease

- Exclusively in girls, normal development for 6-8mo, then regression, handwringing, loss of speech and use of hands. X-linked dominant deletion of MECP2.

Rett Syndrome

- Normal development until age 2 then major loss of verbal, social skills w/ autistic like behavior.

Childhood Disintegrative Disorder

- Lack of mother-child eye contact, language delay/repetitive language, peroccupation w/ “parts of toys” before age 3.

Autism

- Problems with social skills (usually recognized in preschool) w/ reserved verbal ability.

Asperger

A 7 year old boy is brought in by his parents. They report he must be told several times to complete his chores, they cannot get him to focus on completing his homework (he is easily distracted), and that he often loses his shoes, pencils, books, etc.

- Diagnosis? Normal, age appropriate behavior.
- Next best step? Get information from the child's school/teacher
- Risk factors for ADHD? 77% heritability. LBW, tobacco/ETOH exposure
- Comorbid conditions? ODD/CD in 30-50%
- Treatment? Methylphenidate-
  Amphetamine-
  Atomoxetine-
  Randoms-

ADHD Meds

Medication	MOA	Side Effects
Methylphenidate (Concerta, Ritalin)	Blocks DA reuptake	Nausea, ↓appetite, incr HR and BP, stunted growth
Amphetamine (Adderall)	Blocks DA/NE reuptake & stimulates release	
Atomoxetine (Strattera)	NE reuptake inhibitor. Non stimulant	Dry mouth, insomnia, decreased appetite
BP meds (clonidine, guanfacine)	Alpha 2 agonists, reduce peripheral SNS	Decreased BP. Causes sedation
Antidepressants (SNRIs, TCAs, MAOIs)	Prevents NE reuptake and increases in synapse	Dietary restictions w/ MAOI. Arrhythmias in TCAs

A 14 year old boy is sent for court mandated counseling. He stole his neighbor's lawn mower and then set fire to his tool shed. He has a 5 year history of truancy from school and assaulted a 13 year old school mate.

Conduct Disorder. Need sxs for 6mo.
Comorbid substance abuse.
May progress to anti-social personality disorder.

A 14 year old boy is brought in by his grandmother. For the past year, he has been getting in trouble at school for being argumentative and disrespectful to his teachers. He defies the rules she sets for the house and often deliberately annoys her.

Oppositional Defiant Disorder. Need sxs for 12mo.
Stops just short of breaking the lay or physically harming others.

A 9 year old boy is sent to counseling at the recommendation of his teacher. She states that at least once a day he makes loud grunting noises and hand movements that are disruptive to the class.

- Dx? For tics to qualify as Tourettes they must occur at least once a day for 1 year w/o a tic-free period longer than 3mo.
- Comorbid conditions? Look for the compulsions of OCD
- Tx?
  - First line? Clonidine 2/2 relatively benign S/E profile
  - Most Effective? Haloperidol or pimozide- DA-receptor antagonists.

- 7 year old complains of frequent abdominal pain resulting in many missed school days. He never gets the pain on the weekends or in the summer.

  Separation Anxiety Disorder
- 6 year old adopted child is brought in because she has not formed a relationship with her adoptive parents. She is inhibited and hyper vigilant.

  Reactive Attachment Disorder
- An 18mo old baby has recently been regurgitating and re-chewing her food. She had previously been eating normally.

  Rumination Disorder. Check lead levels.
- 6y/o stools in her clothes once every 2 weeks.
  - Next best test? Check for fecal retention.
  - Tx? Behavioral modifcation that only rewards
- 6 y/o urinates in her clothes once a day.
  - Next best test? UA and urine culture.
  - Tx? Alarm and pad for 6wks. TCAs reduce bed wetting but relapse is common. DDAVP has the same prob + S/E = headaches, nausea, and hyponatremia.