

Torey Stories

Compiled using the following resources:
First Aid, 2017
Firecracker

Osteogenesis Imperfecta

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“The Imperfect Bone”



- 1) **Osteogenesis imperfecta**
 - Imperfect bone
- 2) **COL1A1 / COL1A2 gene defects**
 - “COLI” name tag on collar
- 3) **Autosomal dominant**
 - ADd = plus sign
- 4) **Affects type I collagen**
 - 1+1 = type 1
- 5) **“BITE” mnemonic** - see next four points
 - Dog biting the bone
- 6) **Bone = multiple fractures**
 - cracks in bone
- 7) **I (eye) = blue sclera**
 - dog’s blue collar, blue eyes
- 8) **Teeth = dental imperfections**
 - dog’s teeth
- 9) **Ear = hearing loss, abnormal ossicles**
 - ear muffs on dog

Menkes disease

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“Monkeys in a barrel”

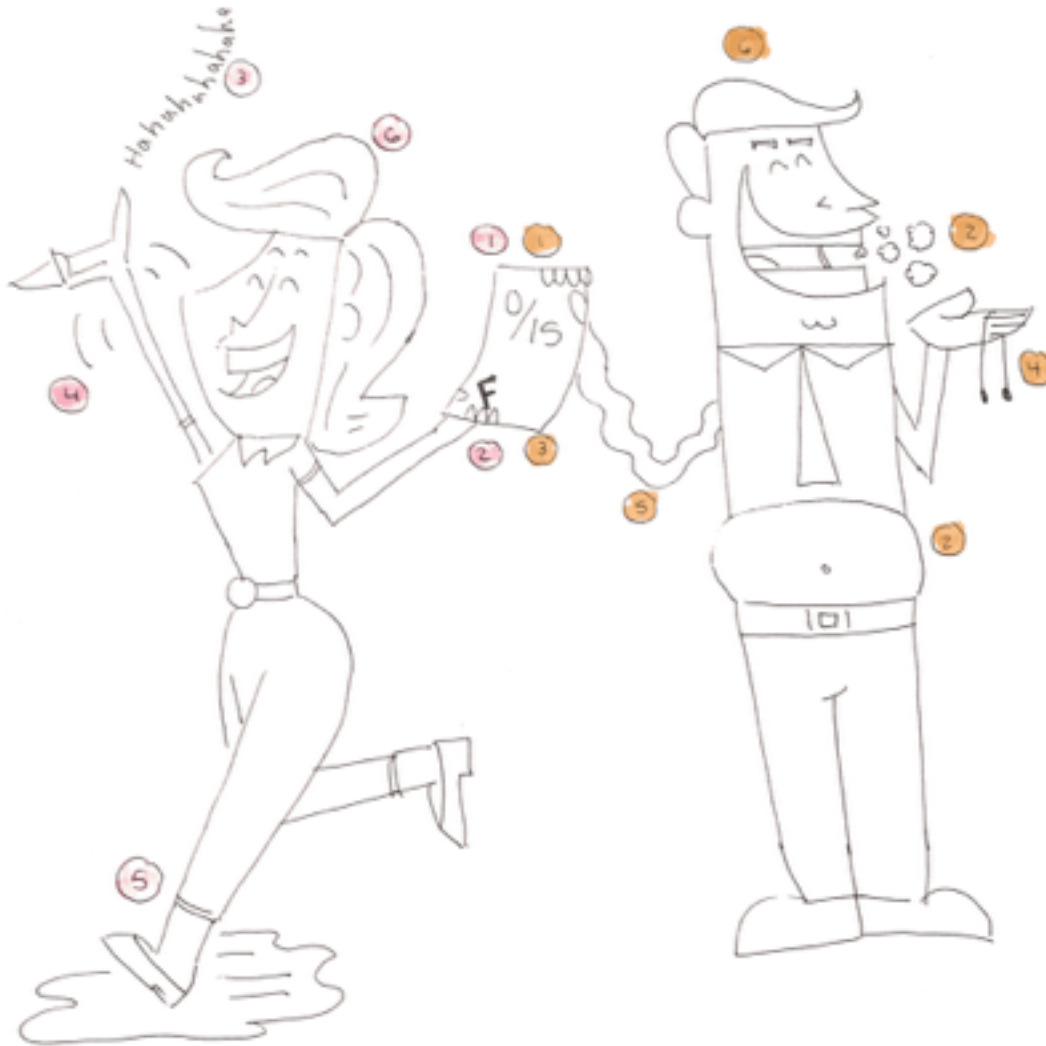


- 1) **Menkes disease**
 - “monkeys” in a barrel
- 2) **X-linked recessive**
 - recess eXit
- 3) **Impaired copper transport** - defective Menkes protein
 - 1 cent sign over monkey barrel
- 4) **Decreased lysyl oxidase activity** - for which copper is a cofactor
 - Lysol spray
- 5) **Brittle, “kinky” hair**
 - kid’s frizzy hair
- 6) **Growth retardation**
 - short kid
- 7) **Hypotonia**
 - kid’s wavy arm

Prader-Willi & Angelman syndrome

“Ma & Pa”

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Prader-Willi syndrome

- 1) **Chromosome 15**
 - 0/15 on the paper
- 2) **Hyperphagia & obesity**
 - Eating food, round belly
- 3) **Intellectual disability**
 - “F” grade on paper
- 4) **Hypogonadism**
 - small beads on string
- 5) **Hypotonia**
 - decreased arm tone
- 6) **Paternal gene imprinted**
 - “Pa” = Paternal gene = Prader-Willi

Angelman syndrome

- 1) **Chromosome 15**
 - 0/15 on the paper
- 2) **Intellectual disability**
 - “F” grade on paper
- 3) **Inappropriate laughter**
 - Hahahahaha
- 4) **Seizures**
 - waving arm
- 5) **Ataxia**
 - slipping on water, losing balance
- 6) **Maternal gene imprinted**
 - “Ma” = Maternal gene = angelMan

Cri-du-chat syndrome

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“Crying Cat”

- 1) High-pitched crying in infant**
 - Meowwww!
- 2) Microcephaly and facial abnormalities - epicanthal fold**
 - Cat’s small head/face
- 3) Intellectual disability**
 - “F” grade on paper
- 4) Ventricular septal defect**
 - heart-shaped name tag with line between ventricles
- 5) Deletion of short arm of chromosome 5**
 - cat’s five, short legs

Williams syndrome

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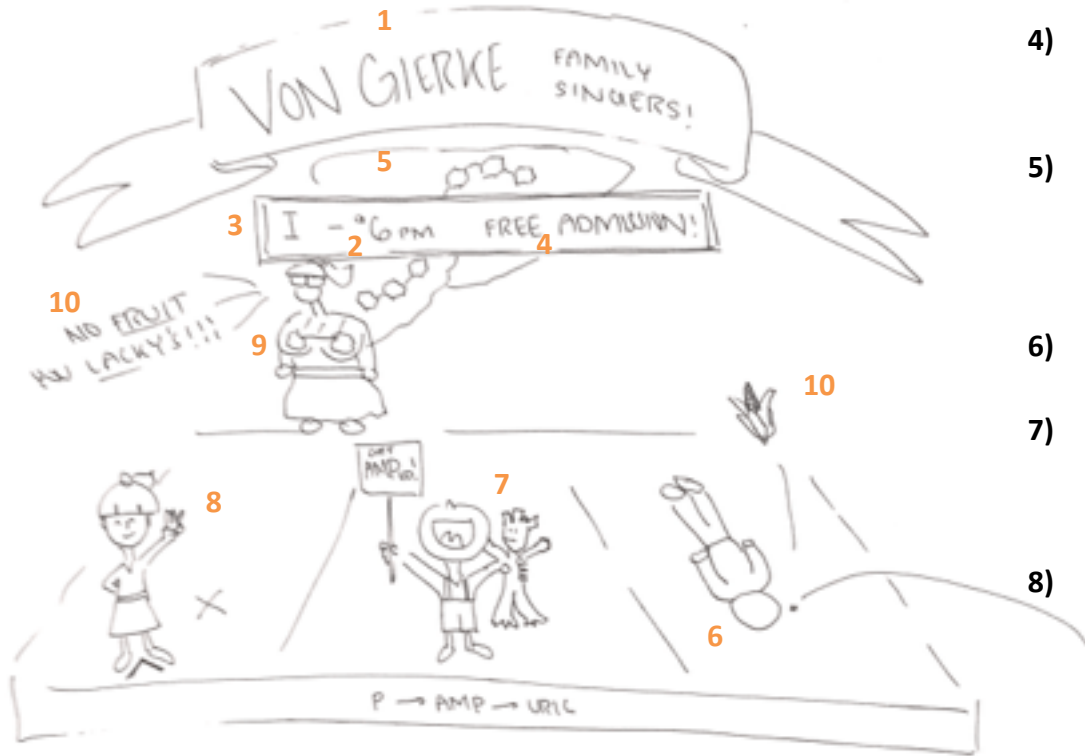


“Will the ice cream guy”

- 1) Deletion of long arm on chromosome 7**
 - man’s long arm, open 24/”7”
- 2) Elastin gene deleted – ELN**
 - eLASTIN’ ice cream
- 3) Elfin features – wide mouth, full lips/cheeks, short/flat nose, wide-spaced teeth**
 - elf ears on man
- 4) Intellectual disability**
 - “F” grade on paper
- 5) High sociability**
 - heart-shaped name tag with line between ventricles
- 6) Hypercalcemia**
 - up arrow on ice cream truck
- 7) Supravalvular aortic stenosis**
 - heart with narrow aortic valve on truck door

Von Gierke disease

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- 1) **Von Gierke disease**
 - "Von Gierke Family Singers"
- 2) **Glucose-6-Phosphatase deficiency**
 - (G)6 PM on sign
- 3) **Type I glycogen storage disease**
 - I - (G)6 PM sign (I = type I)
- 4) **Prevents G6P >> free glucose reaction at liver**
 - "Free Admission" = prevents free glucose
 - results in glycogen accumulation in liver (see 5)
- 5) **Hepatomegaly**
 - Large liver design behind sign with glycogen ribbon on it
 - Increased glycogen accumulation in liver due to lack of free glucose transformation
- 6) **Severe fasting hypoglycemia**
 - Kid on far right passed out from no sugar
- 7) **Gout from hyperuricemia**
 - "Groot" = grout
 - decreased phosphate >> increased AMP (Get amped sign) >> increased uric acid
- 8) **Increased triglycerides - results in xanthomas and increased VLDL**
 - Girl holding up "three" fingers
 - Xanthomas = "X" stage marking
 - VLDL = "V" stage marking
- 9) **Increased blood lactate**
 - Nanny in background lactating through dress
- 10) **Treatment: glucose/cornstarch and avoid fructose and galactose**
 - Corn thrown at boy on right
 - fructose = "fruit" being yelled by nanny
 - galactose = "LACKys" being yelled by nanny

Pompe disease

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1) Pompe disease

- Bastille performing their hit “Pompeii”

2) Lysosomal acid alpha-1,4-glucosidase deficiency

- Lyrics = lysosomes
- April 14th = alpha-1,4

3) Cardiomegaly and hypertrophic cardiomyopathy

- Large heart sign = cardiomegaly
- “my love” = myopathy
- these lead to exercise intolerance

4) Exercise intolerance

- Girl passed out on stage
- Must not confuse with hypoglycemia symbol in the Von Gierke story

5) Hypotonia

- girls loose arm holding liver

6) Liver involvement

- liver sign
- triad = heart, liver, muscle

Cori disease

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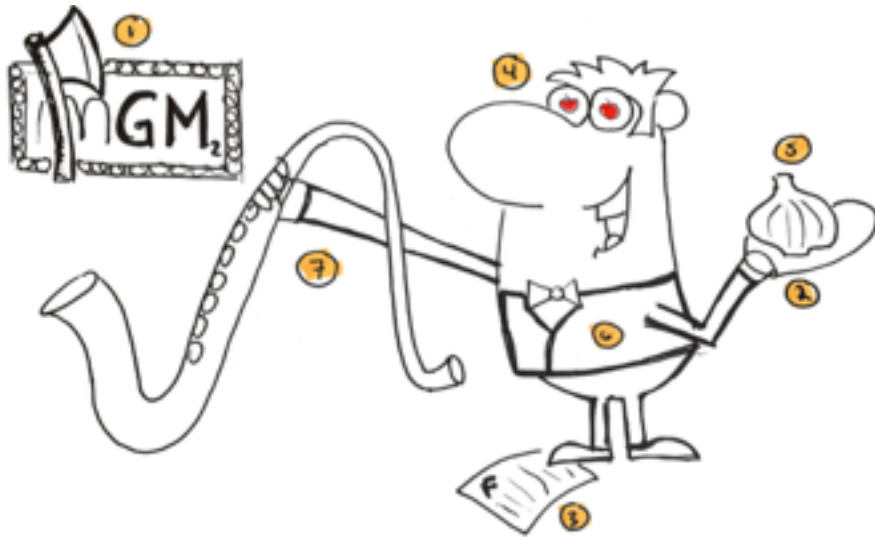
- 1) **Cori disease** = Corgi
- 2) **Accumulation of dextran** = “Dex” the Corgi
- 3) **alpha-1,6-glucosidase deficiency** - branching enzyme
 - branch in Corgi’s mouth
 - April 16th = alpha-1,6
- 4) **Normal blood lactate levels** - compare to Von Gierke disease which has elevated
 - “normal milk” bowl
- 5) **Gluconeogenesis intact**
 - biggest INTACT stick wins
 - abnormal glycogenolysis

Lysosomal Storage Disorders

Compiled using the following resources:
First Aid, 2017
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Tay-Sachs disease

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“A-shaped Saxophone”

- 1) Hexoaminidase A deficiency** - causing accumulation of **GM2 ganglioside**
 - AX = heXoamindase A
 - **MGM(2) sign** = GM2 ganglioside
- 2) Progressive neurodegeneration**
 - oven mitt (not to be confused with peripheral neuropathy as in other sketches)
- 3) Developmental delay**
 - “F” grade on paper
- 4) “Cherry-red” spot on macula**
 - cherries in eyes
- 5) Onion skin lysosomes**
 - “F” grade on paper
- 6) No hepatomegaly** - vs. Niemann-Pick
 - Clean shirt
- 7) Tay-Sachs disease**
 - “A” shaped SAXophone = “tAy-SAX”
 - Ashkenazi jewish descent
 - Exaggerated startle response
 - Death in childhood

*Remember: all lysosomal storage disease are **autosomal recessive** except for Fabry and Hunter

Niemann-Pick disease

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“Pick your nose Newman”

I realize that in the Simpsons this is Ralph... For this sketch, he is Newman



- 1) Niemann-Pick** - deficiency in **sphingomyelinase** causing **sphingomyelin** accumulation
 - “Pick” = Niemann-PICK
 - Pick your nose with your “sphynger” = sphingomyelinase/sphingomyelin
- 2) Progressive neurodegeneration**
 - oven mitt (not peripheral neuropathy as in other sketches)
- 3) Foam cells** - aka lipid-laden macrophages
 - Foam on the ground
 - “lipid belt” = lipid laden macrophages
- 4) “Cherry-red” spot on macula**
 - cherries in eyes

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Fabry disease

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"Fabry Fairy"

- 1) alpha-galactosidase A deficiency**
 - GOLly = GAL = GALactosidase
 - "As" on wings = alpha, A
- 2) Ceramide trihexoside accumulation**
 - "three hexes" = ceramide TRIHEXoside
- 3) Peripheral neuropathy**
 - gloves on hands
- 4) Angiokeratoma - cutaneous lesions of capillaries**
 - bottle CAPs = capillaries
- 5) Hypohydrosis - diminished sweating**
 - # on face
- 6) Late: progressive renal failure**
 - kidney belt
- 7) Late: cardiovascular disease**
 - heart on chest
- 8) X-linked recessive**
 - Recess eXit

*Remember: all lysosomal storage disease are **autosomal recessive** except for Fabry and Hunter

Metachromatic Leukodystrophy

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“Something is off with Master Luke”

- 1) Arylsulfatase A deficiency** - leading to accumulation of **cerebroside sulfate**
 - Luke watching “The Little Maid” (Ariel = ARYLsulfatase) and thinking he’s a mermaid
- 2) Metachromatic leukodystrophy**
 - LUKE Skywalker thinks he’s a multi-colored mermaid = METACHROMatic LEUKodystrophy
- 3) Central and peripheral neuropathy**
 - oven mitt on head = central
 - oven mitts on hands = peripheral
- 4) Ataxia**
 - stack falling over in his hand, similar to Sketchy’s representation of ataxia
- 5) Dementia**
 - Think of the story here... Luke Skywalker thinks he’s a mermaid = Dementia (kind of stretch but whatever)

*Remember: all lysosomal storage disease are **autosomal recessive** except for Fabry and Hunter

Gaucher & Krabbe disease

"Grouchy Grandpa and his Krab"

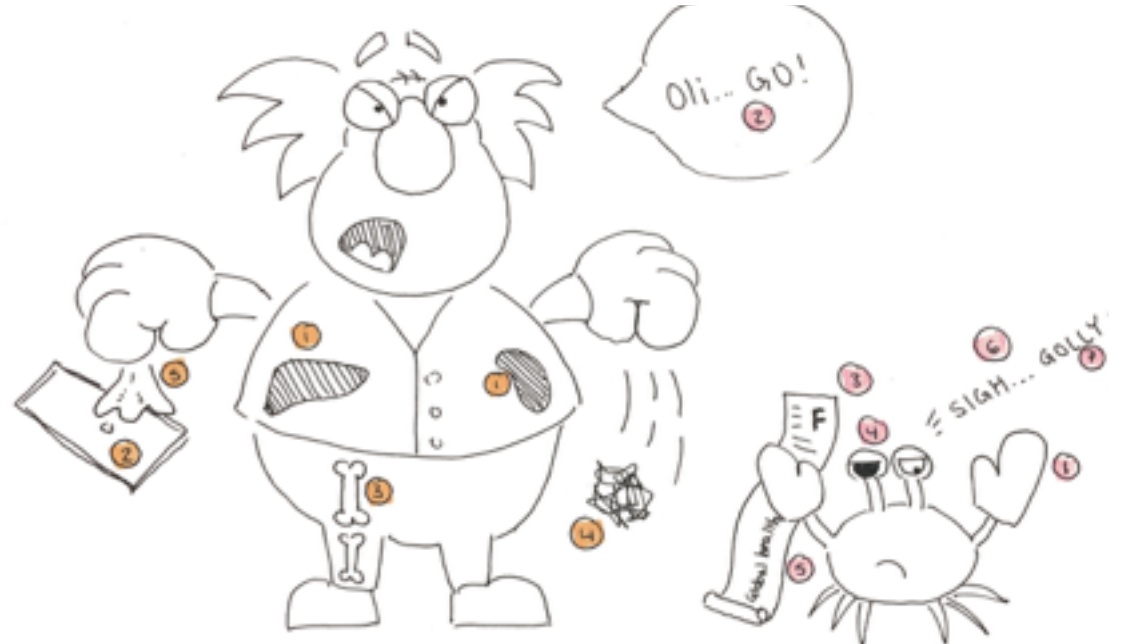
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Gaucher disease

- 1) Hepatosplenomegaly**
 - liver/spleen stain on shirt
- 2) Pancytopenia**
 - pan stuck to mans hand
- 3) Bone involvement -**
osteoporosis, avascular necrosis of femur, bone crisis
 - bones on man's pants
- 4) Gaucher cells -** lipid-laden macrophages resembling crumpled tissue paper
 - crumpled paper
- 5) Glucocerebrosidase deficiency resulting in glucocerebroside accumulation -** alternatively, beta-glucosidase
 - Glue on hand = GLUcocerebrosidase
 - β glucosidase = β ad grandpa

Krabbe disease

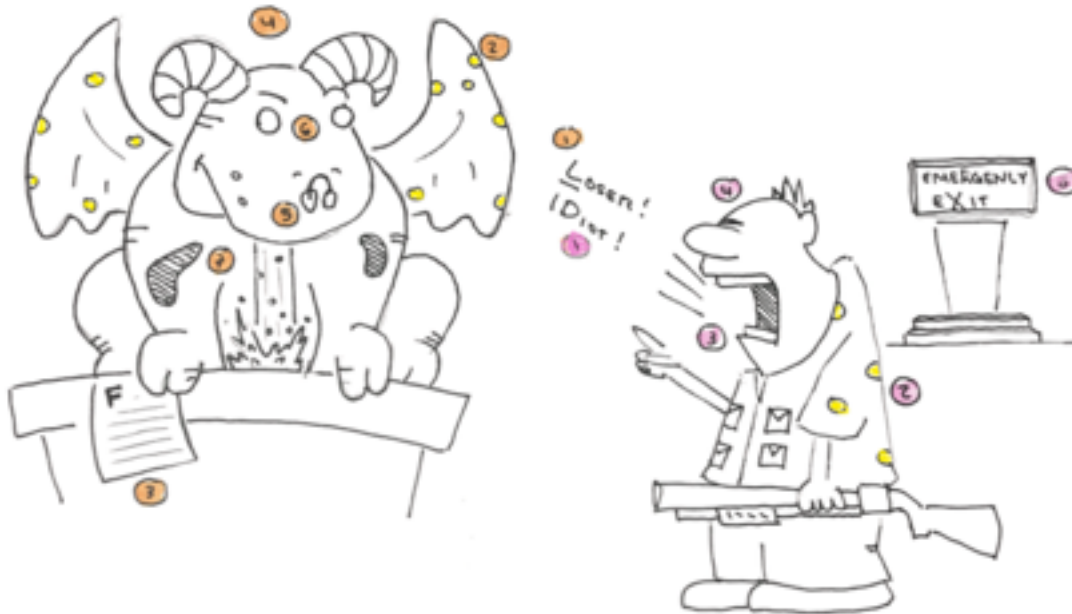
- 1) Peripheral neuropathy** oven mitts on crab
- 2) Oligodendrocyte destruction** "Oli... GO!" = OLIGOdendrocytes
- 3) Developmental delay** "F" grade on paper
- 4) Optic atrophy** crab's black eye
- 5) Globoid cells** "global health" on paper
- 6) Psychosine and galactocerebroside accumulation** "sigh" - sigh-chosine = psychosine
- 7) Galactocerebrosidase deficiency** "golly" = GALLY = GALactocerebroside
 - Galactocerebroside is an important component of myelin
 - Death usually occurs before 2 years of age



- **No weakness:**
 - Think: **Gaucher** or **Fabry's**
- **Weakness**
 - in baby — with cherry red spot (**Krabbe's**)
 - in baby — without cherry red spot (**Tay-Sachs**)
 - Ataxia ~2 years (**Metachromatic leukodystrophy**)
 - Older child with hepatosplenomegaly (**Niemann-Pick**)

Hunter & Hurler syndrome

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Hurler syndrome “Hurling Gargoyle”

- 1) **alpha-L-Iduronidase deficiency**
 - “Loser IDiot”
- 2) **Heparin sulfate & dermatan sulfate accumulation**
 - yellow spots on gargoyle
- 3) **Developmental delay**
 - “F” grade on paper
- 4) **Gargoylism**
 - gargoyle (coarse facial features)
- 5) **Airway obstruction**
 - nose plugs on gargoyle
- 6) **Corneal clouding**
 - blank eyes of gargoyle
- 7) **Hepatosplenomegaly**
 - liver-spleen shaped spots on gargoyle

Hunter syndrome “Angry Hunter”

- 1) **Iduronate sulfatase deficiency**
 - “IDiot”
- 2) **Heparin sulfate & dermatan sulfate accumulation**
 - yellow spots on hunter
- 3) **Aggressive behavior**
 - Yelling at gargoyle
- 4) **No corneal clouding (as seen in Hurler’s)**
- 5) **Autosomal recessive**
 - emeRgency eXit

- **I-Cell Disease**

- failure of processing in **Golgi Apparatus**
- no addition of **Mannose-6-Phosphate** (thus, enzymes aren't directed to lysosome and are secreted out of cell)
 - labs: increased extracellular enzymes, decreased intracellular enzymes
- see **intracellular inclusions** seen in fibroblasts/WBCs

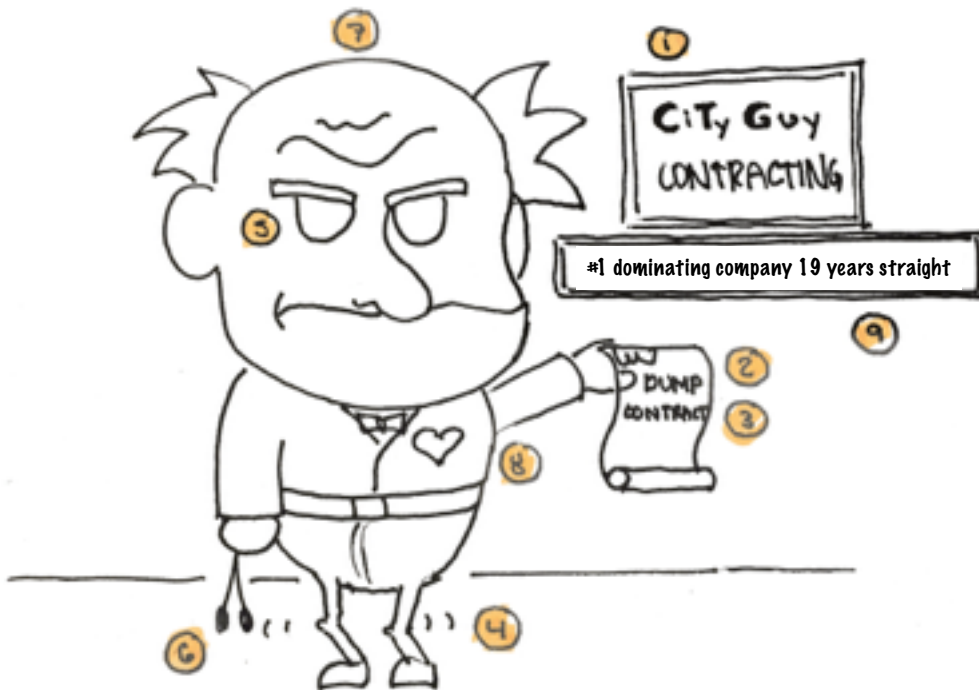
- **Pompe disease**

- both **glycogen storage** and **lysosomal storage** disease
- alpha-1,4-glucosidase deficiency
- glycogen accumulates in lysosomes
- death in infancy/childhood

Myotonic Type I muscular dystrophy

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“Monotone CiTy Guy Contracting Company”



- 1) **CTG trinucleotide repeat**
 - CiTy Guy Contracting company
- 2) **DMPK gene >> abnormal myotonin protien kinase**
 - DuMP Kontract
- 3) **Myotonia** - constant contraction
 - Contract (city guy and dump)
- 4) **Muscle wasting**
 - small, shaky legs
- 5) **Cataracts**
 - cloudy eyes
- 6) **Testicular atrophy**
 - small beads on string in guy's hand
- 7) **Frontal balding**
 - CiTy Guy is balding
- 8) **Arrhythmia**
 - “Heart our Customer’s” pin on guy
- 9) **Autosomal dominant, chromosome 19**
 - #1 dominating company
 - 19 years straight = chromosome 19

Fragile X syndrome

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"Fragile X Farmer dominated the competition"

- 1) **Fragile X syndrome** eXtremely FRAGILE
- 2) **CGG trinucleotide repeat on FMR1 gene**
County Gold Games = CGG, Farmer = FMR
- 3) **Intellectual disability** "F" grade on paper
- 4) **Autism** (above)
- 5) **Macroorchidism** large watermelons = large testicles
- 6) **Long face, large jaw** farmer's long, large jaw
- 7) **Large everted ears** farmer's large ears
- 8) **Mitral valve prolapse** Most Valuable Pharmer = MVP

