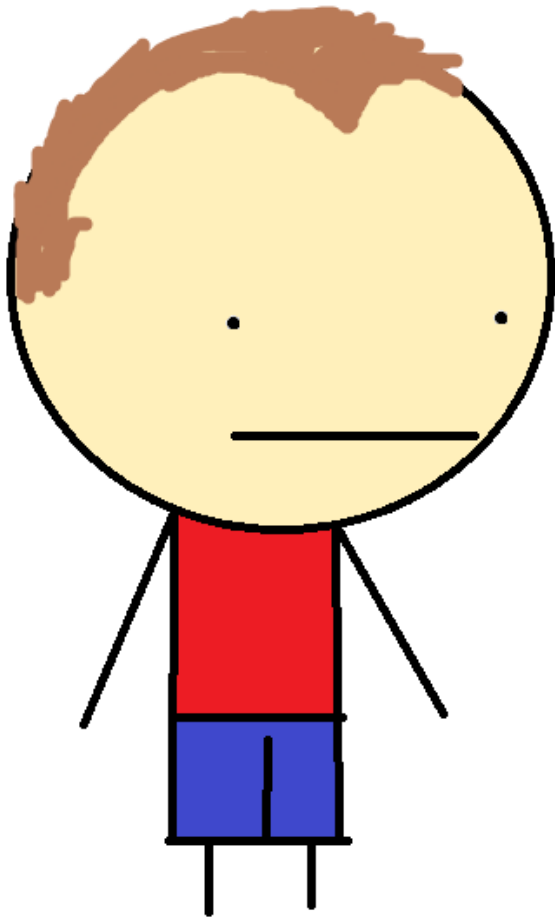
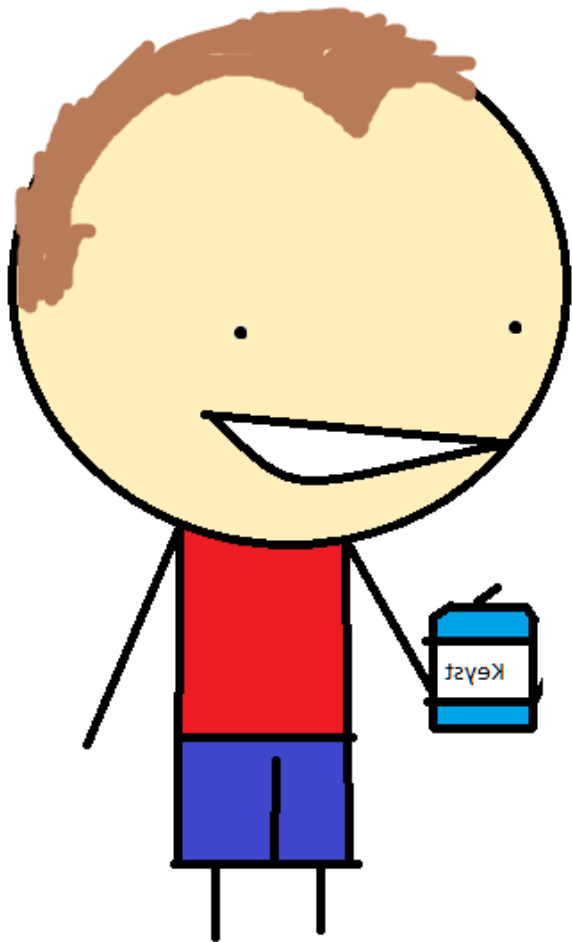


Meet Paul



Paul, despite his characteristics of early male pattern balding, appears to be like any other 30 year old male.

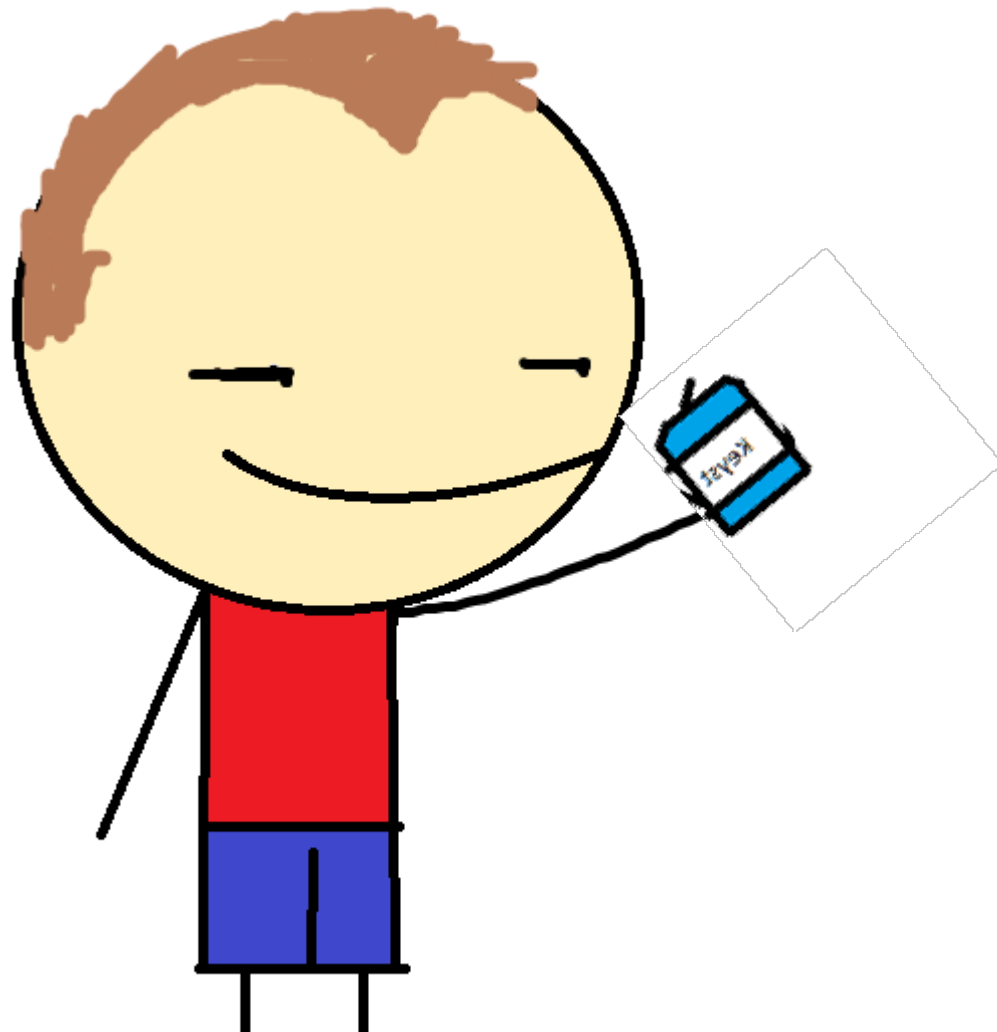
Paul just got home from work and now wants to enjoy a nice cold drink.

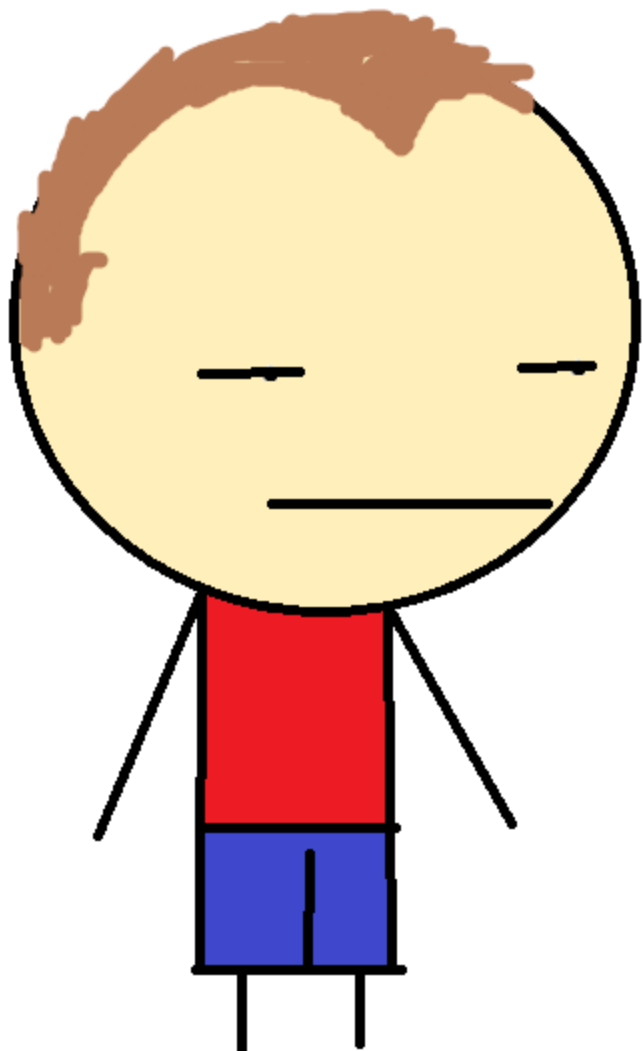


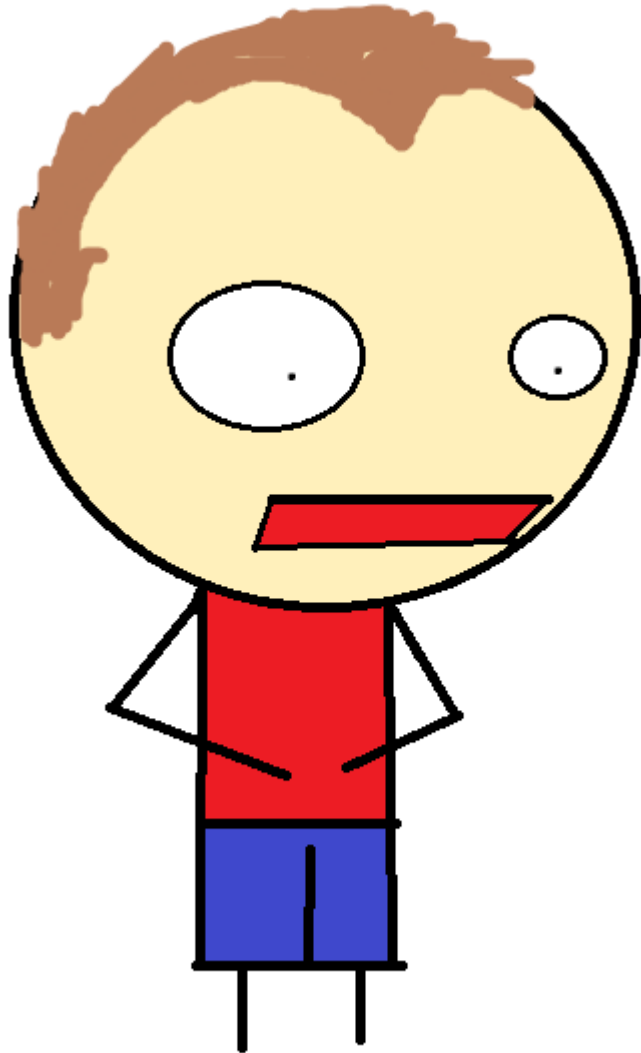




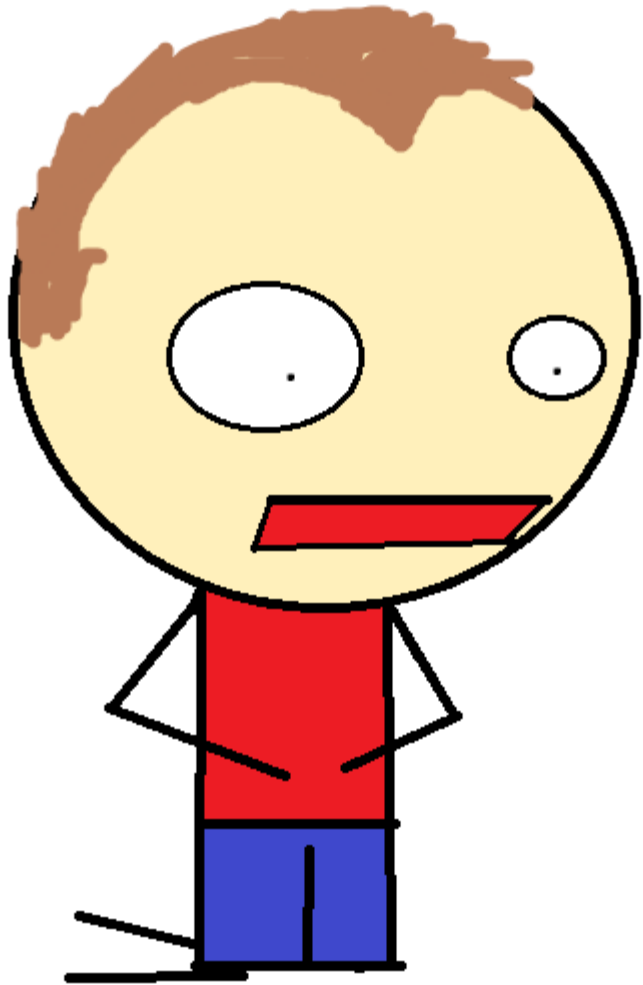


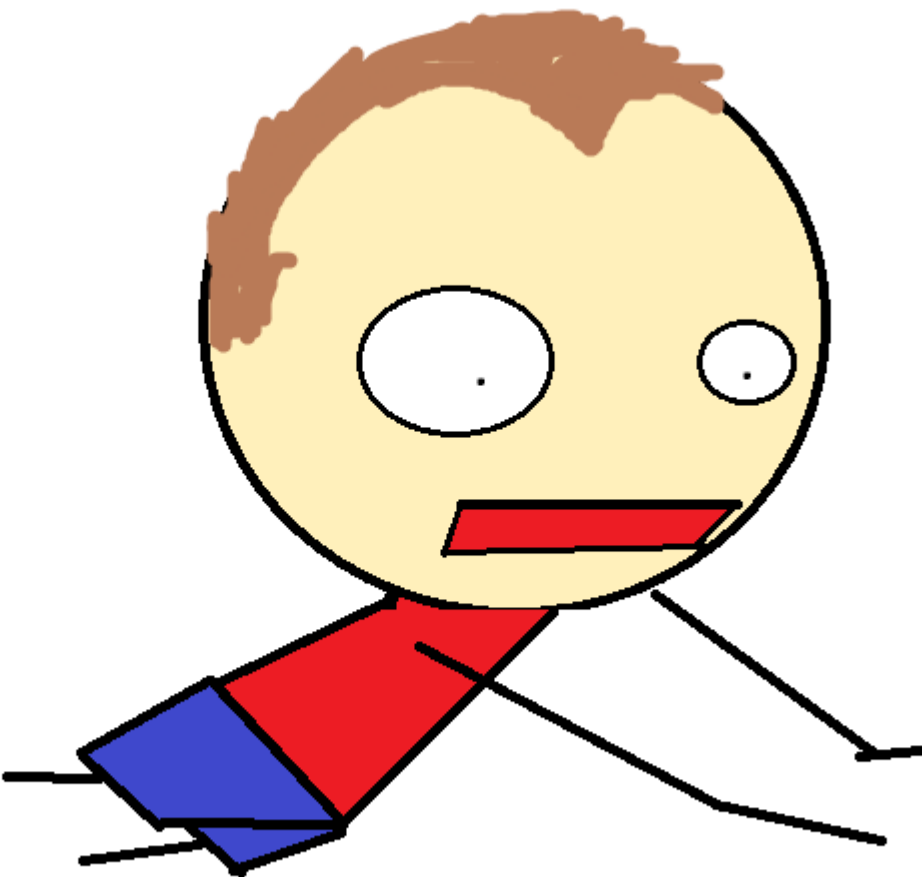




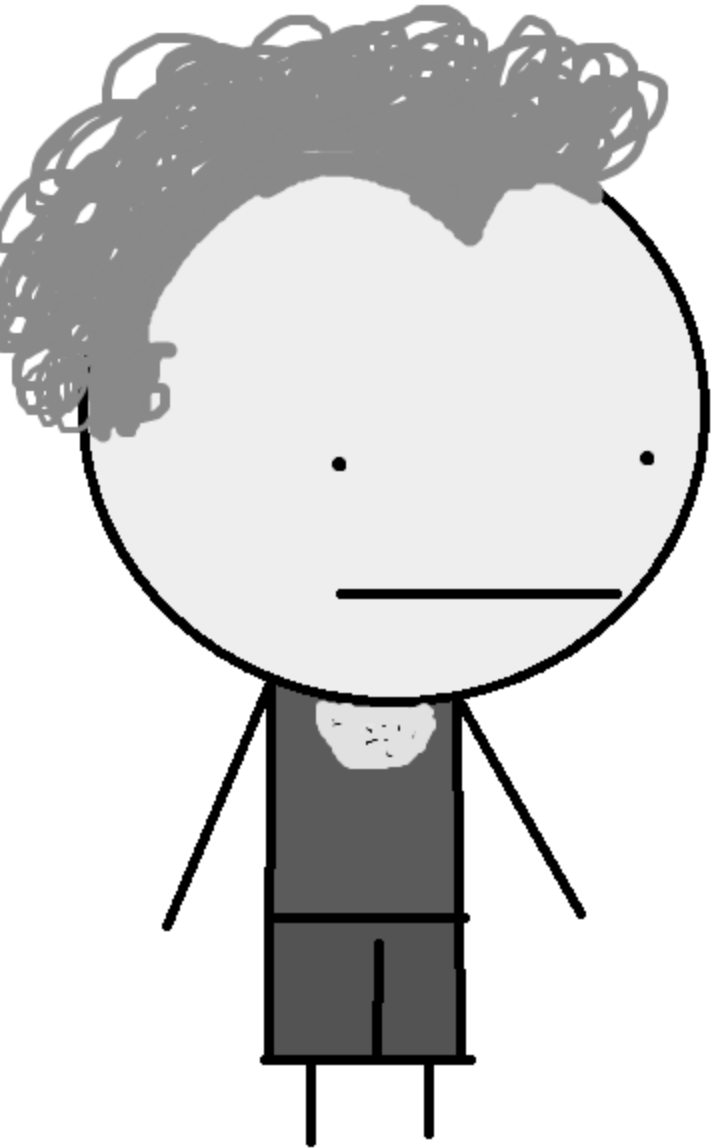


What's this? It appears Paul is experiencing some extreme type of pain.





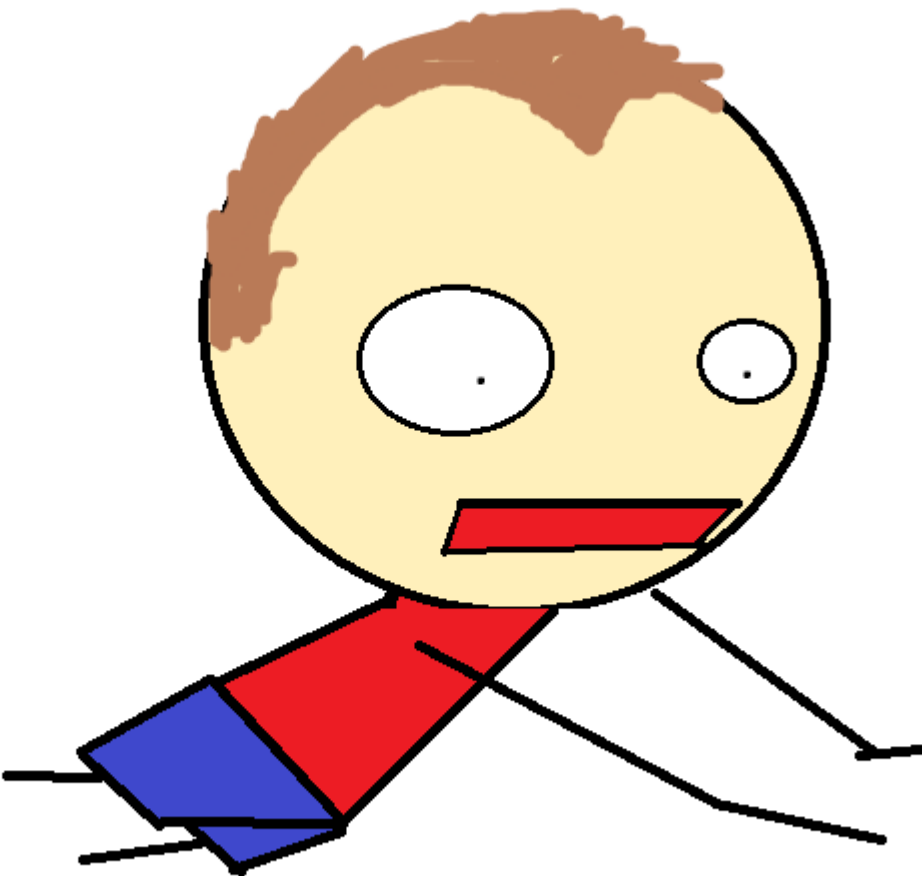
Paul begins questioning where is this pain coming from? He has never felt this way before. Could it be the drink?



Meet Paul age 21







Nope, not the alcohol.

Then what is wrong
with Paul?

Polycystic Kidney Disease

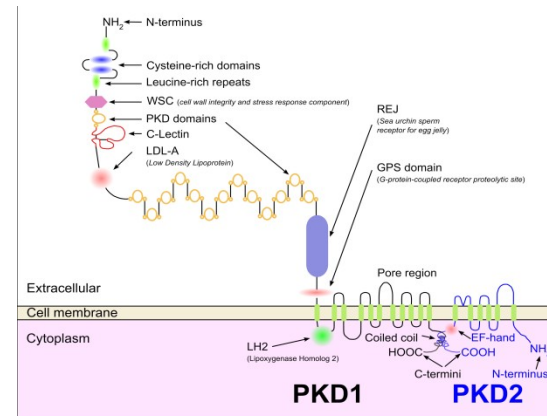
Taurean Butler

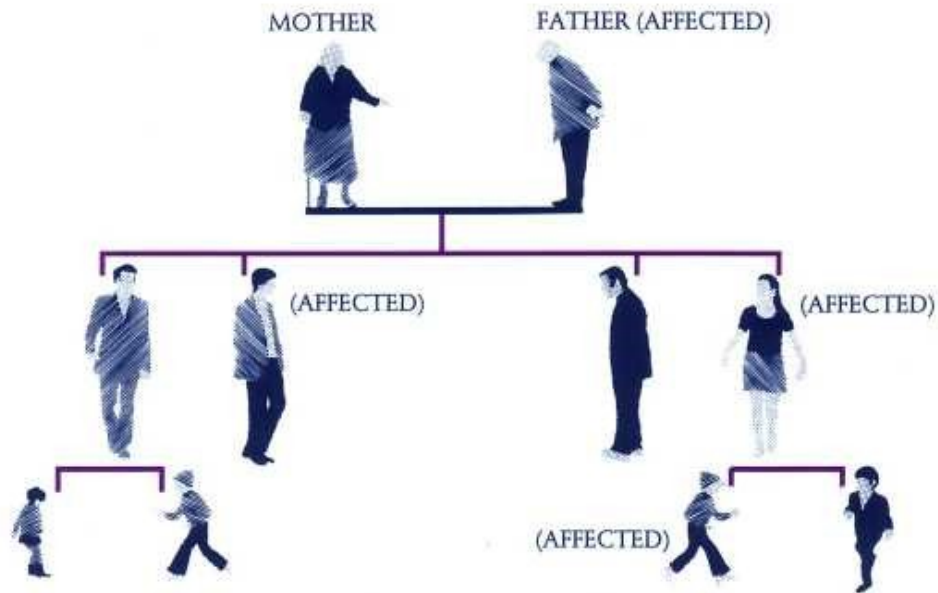
Genomics and Medicine

- Most common genetic life threatening disease
 - 1/400 to 1/1000
- Two Forms of PKD:
 - Dominant Form: Autosomal Dominant Polycystic Kidney Disease (ADPKD)
 - Recessive Form: Autosomal Recessive Polycystic Kidney Disease (ARPKD)

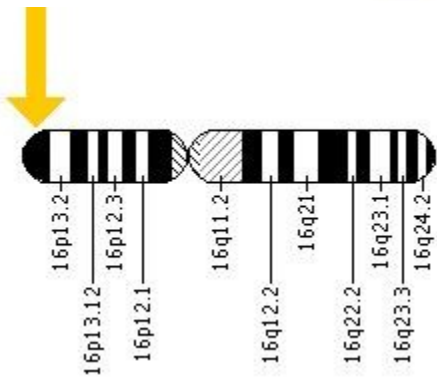
ADPKD

- Far more prevalent of the two
- Can be caused by one of two genes:
 - PKD1
 - PKD2
- Genes responsible for the production of Polycystin-1/2
- Polycystin-glycoprotein that serves in cell/matrix interaction
- 16p13.3-p13.12

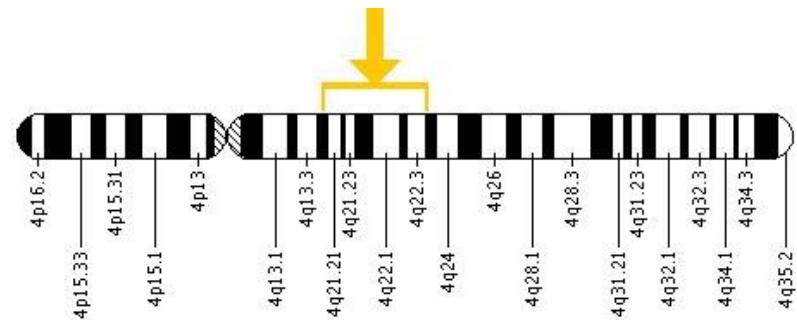




AN EXAMPLE OF HOW ADPKD IS INHERITED



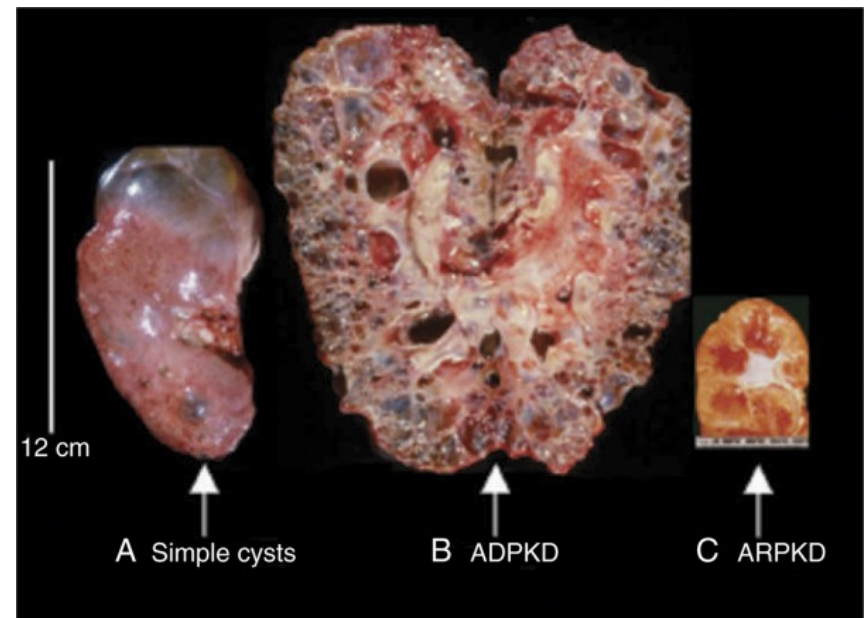
PKD1



PKD2

Symptoms of ADPKD

- Renal Pain
- Bilateral or unilateral renal cysts
- Formation of cysts in other organs
- Kidney enlargement
- Hypertension
- Signs become apparent near adulthood



Characteristics of ADPKD

- Progresses over life
 - 30: 2 cysts
 - 59+: 4 cysts
- End-stage Renal Disease by age 60
- Patients typically heterozygous
 - Homozygous results in early deaths
- Gene mutation affects severity of the disease

Diagnosing the Disease

- Kidney Imaging most prevalent means
- Gene Testing
 - PKD1: Sequence analysis
 - PKD2: Sequence analysis, linkage analysis
- Rare prenatal testing



Treatment

- Primary treatment focuses on reducing morbidity of the disease
 - Ace inhibitors, Flank pain
- Cyst decompression
 - Cyst aspiration, sclerosis agents, surgery
- Preventative Treatments
 - Avoiding: caffeine, smoking, estrogen use

Works Cited

- <http://ghr.nlm.nih.gov/condition/polycystic-kidney-disease>
- <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?l=bookshelf&book=NBK1382>
- <http://www.ncbi.nlm.nih.gov/omim/601313>
- <http://www.ncbi.nlm.nih.gov/omim/263200>