- **Adult Polycystic Kidney Disease (PCKD)** (refer below)
- **Simple Solitary Cyst** (seen in 50% of pts >50yo, asymptomatic but some may undergo malignant degeneration esp if hemorrhagic cyst, can be solitary/multiple uni/bilateral, on surface, no treatment necessary unless infection/tumor present)
- **Acquired Cyst** (develop after long term dialysis)
- **Medullary Sponge Kidney** (ectasia of collecting duct forming cyst-like structures in renal papilla, most are asymptomatic but sometimes hematuria, Type I RTA, recurrent UTIs, nephrolithiasis/hypercalciuria and defects in concentrating ability but NO renal failure, unlike PCKD kidneys are small, good prognosis)
- **Nephronophthisis aka Medullary Cystic Kidney aka Hereditary Tubulointerstitial Kidney Disease** (? of collecting duct forming cysts at the cortico-medullary jxn, eventually there is tubular atrophy, interstitial inflammation, and scarring w/ ESRFD, UA is normal!!!, uniquely there is a defect in concentrating ability such that pts present w/ polyuria, polydipsia, nocturia followed by progressive renal failure by 20yo, unlike PCKD kidneys are small, in addition to retinitis pigmentosa, bad prognosis)

	Autosomal Dominant PolyCystic Kidney/Liver Disease (ADPCKLD) (AD, 1:600)	**Infantile Polycystic Kidney Disease (AR, 1:40,000)**
Genetics	• PKD1 gene mutation (85%) on Chr-p16 (Polycystin-1 = membrane protein found in epithelial of ducts of various organs and is involved in cell-cell and cell-matrix interactions) • PKD2 gene mutation (15%) on Chr-q14 (Polycystin-2 = protein involved in Ca++ signaling)	• PKHD1 gene mutation on Chr6 (Fibrocystin = cell surface receptor important on CD of kidney)
Pathophysiology	abnormal growth of tubule cells → cells proliferate and secrete fluid → bilateral cysts	
	Rough Surface w/ Large Cortical Cysts	Smooth Surface w/ Small Medullary Cysts
Renal Symptoms	• HTN (typically the first Sx) • Progressive RF w/ 50% reaching ESRD at 65yo (4th leading cause of ESRD) • Flank/Back Pain i. Acute: pain occurs w/ cyst rupture, cyst hemorrhage, cyst infection, obstructive uropathy ii. Chronic: gradual enlargement of cysts • Ab Mass • Gross Hematuria from cyst rupture • Polyuria/Nocturia b/c Impaired Concentrating Ability • UTI, Cyst Infection, Pyelonephritis • Nephrolithiasis • Renal Cell Carcinoma	
Extra-Renal Symptoms	(1) Cerebral/Thoracic/Abdominal Aneurysms (2) Colonic Diverticulae and Abdominal Hernias (3) MR/AI (4) Hepatic/Pancreatic/Seminal Vesicle/Arachnoid Cysts a. Suggestive if >4 cysts b. Liver cyst can be quite large causing hepatomegaly, ab pain, early satiety, c. LFTs are usually remarkably normal!!! d. Complications: rupture, infection, compression of bile ducts and veins e. more common in women and women tend to have worse dz 2/2 hormone effects, cyst do not appear till puberty	(1) Symptoms of Fetal Renal Insufficiency (oligohydraminos, Potter's syndrome, pulmonary hypoplasia) (2) Hepatic Fibrosis causing portal HTN
Prognosis	• Normal GFR until ~50yo (even though cysts are present at birth) with slow steady progressive decline to ESRD over 10yrs (4th leading cause) • Common causes of death include HTN, infection, ruptured aneurysm	• Begin to develop renal insufficiency at birth with ESRD during infancy
Treatment	• Surveillance MRA Head • Fenestration of cysts • Liver/Kidney Transplants • Avoid caffeine which has been associated w/ increased cyst formation!!! • NO treatment just Nephrectomy followed by Dialysis/Transplant • There is some evidence that inhibition of RAAS system is helpful • There is some evidence that decrease in cAMP slows progression of disease w/ somatostatin analogues • Sirolumus is found to be effective • Drain cysts if symptomatic (chronic pain or infection) but has not been found to slow rate of GFR decline	