FETAL RENAL CYSTIC DISEASES

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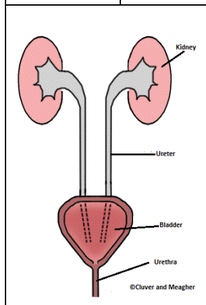
Normal		Autosomal recessive polycystic kidney disease (ARPKD): Potter I	Multicystic kidney disease (MCKD): Potter II	Autosomal dominant polycystic kidney disease (ADPKD): Potter III	Hydronephrosis: Potter IV	Hyperechoic kidneys + Genetic Syndromes/ aneuploidies	Unexplained Hyperechoic kidneys
Ultrasound Finding	**Kidneys**	• Bilateral, hyperechoic enlarged kidneys from 16w • Diagnosis only made in 50% at mid trimester • Renal length most valuable in diagnosis • 1-2 mm non-communicating • Renal cysts, some may have macrocystic disease • Cystic fusiform dilatation of the collecting ducts	• Often unilateral (80%) • Large hyperechoic kidney • Multiple and non-communicating cysts of variable shapes and sizes • Loss of reniform shape • Ultrasound findings after 14 weeks	US findings after 14-16 weeks • Bilateral enlarged or grossly enlarged kidneys • Hyperechoic cortex only • Exaggerated corticomedullary differentiation • Cysts in pancreas, liver, spleen, and CNS • No cysts seen in kidneys	• Caliectasis: connections visible between calyces and pelvis • Parenchyma not echogenic • 'Eggshell sign': crescent of increased echogenicity at caliceal/ parenchymal interface • Unilateral or bilateral • Associated with megaureter and perinephric urinoma • Present if renal pelvis AP diameter >5mm at less than 32 weeks and >10mm after 32 weeks	Echogenic kidneys May be enlarged Associated with other anomalies	• Unilateral or bilateral • No other anomalies
	Bladder	Absent	Normal	Normal	Absent/normal/ dilated/thick-walled bladder	Normal	Normal
	AFI	Develop severe oligohydramnios	Normal if unilateral, oligohydramnios if bilateral	Usually normal	Normal or oligohydramnios (dependent on cause)	Normal/reduced	Normal



1st Trimester

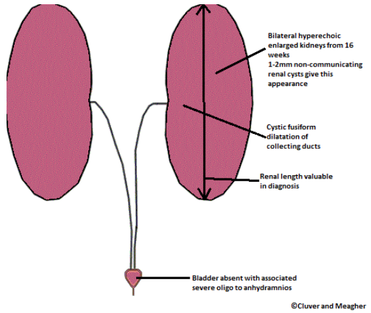
http://youtu.be/EQg2hela7g
http://youtu.be/56x9LmRJVM0
http://youtu.be/4y7OjAq84a0

2nd Trimester

http://youtu.be/ioUX4zQyL5U
http://youtu.be/3Nco-BASv1Y
http://youtu.be/Vae5cb2JP6M

3rd Trimester

http://youtu.be/s0TbTLOCIVk

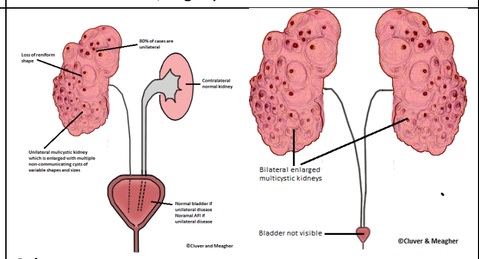


ARPKD

Case 1
http://youtu.be/I-zLC9nWdD8
http://youtu.be/5bLKkZJn8HY

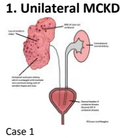
Case 2
http://youtu.be/fbVHTNYctf0

2 weeks later:
http://youtu.be/Ru9Flisd3iA



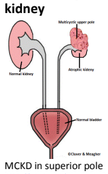
Subtypes

1. Unilateral MCKD



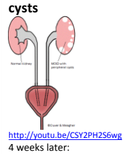
Case 1
http://youtu.be/gEteOD-8mzI
http://youtu.be/ViHL-6ZSkXI
Case 2
http://youtu.be/6d51hTe3k_Y
http://youtu.be/pXabNkN7jO
http://youtu.be/tKGz5bkX6Pc
http://youtu.be/SUi-w8Zloow

2. Unilateral MCKD in a small atrophic kidney



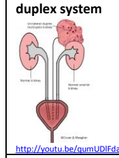
MCKD in superior pole
http://youtu.be/IWwTHrwPKbI

3. Unilateral MCDK presenting with simple peripheral cysts



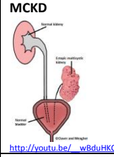
http://youtu.be/CSY2PH2S6wg
4 weeks later:
http://youtu.be/aO_NbALLiYs

4. Unilateral MCKD in one moeity of a duplex system



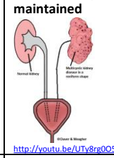
http://youtu.be/qumUDIFdgwc

5. Unilateral pelvic ectopic kidney with MCKD



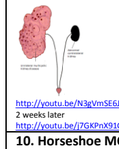
http://youtu.be/__wBduHKQck

6. Unilateral MCKD reniform shape maintained



http://youtu.be/UTv8rg0O51M

7. Unilateral MCKD with an abnormal contralateral kidney

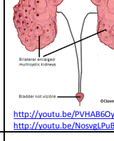


http://youtu.be/N3gVmSE6JhI
2 weeks later
http://youtu.be/J7GKPnX91GE

8. Unilateral MCKD with contralateral renal aplasia)

http://youtu.be/r8oLEZOiBO8

9. Bilateral MCKD



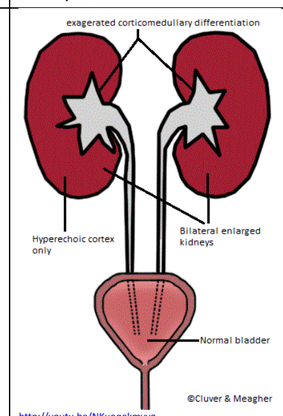
http://youtu.be/PVHAB6OyfoA
http://youtu.be/NosvgLPu8DA

10. Horseshoe MCKD

http://youtu.be/uzOMilcY3pQ
http://youtu.be/TL5vyfuJ0k

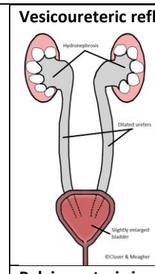
11. Bilateral MCKD: Unusual presentation

http://youtu.be/94bUyDRy1Nc
http://youtu.be/NRNHspSthc0



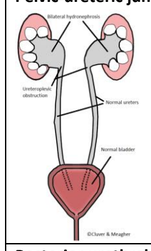
http://youtu.be/NKuoqekmvvg
http://youtu.be/Kz2nTj02hy4
http://youtu.be/V8ArP8sGvaA
http://youtu.be/5keyegsYj_s

Vesicoureteric reflux / obstruction



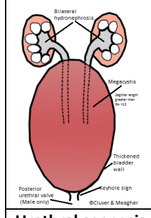
http://youtu.be/j74CvWUTtGI
http://youtu.be/4sT0BPaZv84
http://youtu.be/TcfuHMNqqPA
http://youtu.be/oxoFVXGy0Jk
http://youtu.be/qD_QXmCu7Pk
http://youtu.be/RSXgvWIAiGQ
http://youtu.be/VC6rur0JKBM
http://youtu.be/Gc2jP4TT9ag
http://youtu.be/N89BGCH4uVA
http://youtu.be/ZTF5qO8q--8\

Pelvic-ureteric junction obstruction (PUJ)



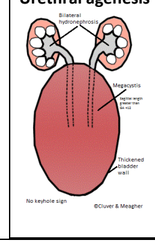
http://youtu.be/VNAktIZS0pM

Posterior urethral valves

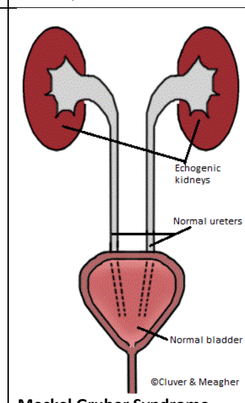


1st trimester
http://youtu.be/-dSDKow2JEM
http://youtu.be/mCDMQSM7yWo
http://youtu.be/ia8vY8tSwTU
http://youtu.be/uAW9FRMHUus

Urethral agenesis



1st trimester
http://youtu.be/7WQEpD81YcM
2nd trimester
http://youtu.be/U4RVvdm5NrA
http://youtu.be/OzVQYjkEJTw



Meckel Gruber Syndrome

Case 1
http://youtu.be/zXd48KpJ9oY
http://youtu.be/ytNIeeHXOL0
http://youtu.be/YA81I1TaYao
http://youtu.be/VOtqpBc_zQ4

Case 2
http://youtu.be/L0twMtDa4FM
http://youtu.be/F_KeD27DUY4
http://youtu.be/katC_ww7j8Y

Trisomy 18

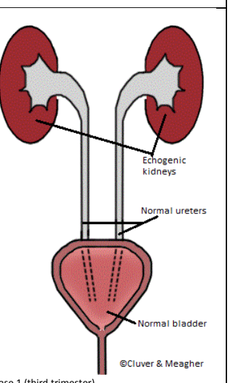
http://youtu.be/HnzKlyWmNII
http://youtu.be/GIG4zEWHn7M

Trisomy 13

http://youtu.be/RgHDugGR-BY
http://youtu.be/5iVsuObnXqQ
http://youtu.be/29xUFITZcGo

Bardet-Beidl Syndrome

http://youtu.be/DOJ8df_tyvY
http://youtu.be/YDXm59DVe34
http://youtu.be/nIH_qybQQ_A



Case 1 (third trimester)
http://youtu.be/rOxYjO6tu0o

Case 2 (second trimester)
http://youtu.be/JJgsOz_hTgE

	Autosomal recessive polycystic kidney disease (ARPKD): Potter I	Multicystic kidney disease (MCKD): Potter II	Autosomal dominant polycystic kidney disease (ADPKD): Potter III	Hydronephrosis: Potter IV	Hyperechoic kidneys + Genetic Syndromes/ aneuploidies	Unexplained Hyperechoic kidneys
Incidence	1:20 000-40 000	Unilateral 1: 3 000-5 000, Bilateral: 1: 10 000	1: 800-1 000	Most common uropathy 1: 100	Sporadic/genetic-linked/family-linked/age-linked	10-21% hyperechoic kidneys
Associated anomalies	• Potter sequence develops after 20 weeks (oligohydramnios and pulmonary hypoplasia) • Liver cysts, hepatic fibrosis, portal hypertension and biliary duct hypoplasia	• In 30–50% the other kidney is dysplastic or affected by vesico-ureteric reflux • Unilateral: 26% extra-renal anomalies • Bilateral: 67% extra-renal anomalies • Extra-renal anomalies: cardiac (majority), GIT, CNS, spine, single umbilical artery, genital anomalies. • Syndromes: VACTERL, Cerebrodigital, Brachio-oto-renal, Meckel–Gruber, Bardet–Biedl, Fraser, CHARGE	• Not associated with aneuploidies • Polycystic disease of liver, spleen and pancreas rarely seen in the fetus but common in adults	• Aneuploidies: 0,4-1,6% (50% being T21) • Syndromes: 5-10% (VACTERL, Cerebro-digital syndrome, tuberous sclerosis)	Extra-renal: • Cardiac • Neurological • Facial (clefts, cataracts, macroglossia) • Spinal and limb • Diaphragmatic hernia • GIT: exomphalos • Genital and urinary tract anomalies	None
Genetic associations/ Aetiology	• Autosomal recessive • Significant genetic heterogeneity • Single gene disorder: 80% mutation in PKHD1 Gene on chr 6P21 • Can be associated with other single gene anomalies and aneuploidies •4 types: prenatal, neonatal, infantile and juvenile	• 90% non-genetic- linked • 10% autosomal dominant • Monogenic causes: mutations in individual genes, such as TCF2, PAX2 and uroplakins • Heterozygote mutations in several genes Risk of chromosomal anomalies • Unilateral dysplasia: 2-4% • Bilateral dysplasia: 15-18% (Trisomy 13 and 18) or inherited conditions (3-10%) Risk of non-chromosomal syndromes • 5-10%	• Autosomal dominant (90%) • 10%: no family history of ADPKD (genetic mutation) • PKD1 mutations (85%): gene on chromosome 16p13 • PKD2 mutations (15%): gene on chromosome 4q21-q23 • Can be a component of tuberous sclerosis: gene TSC2 on chromosome 16	• Usually not genetic linked <u>Obstructive</u>: • Ureteropelvic junction obstruction (UPJ) 10-30% • Ureterovesical obstruction (UVJ)/megaureter 5-10% • Posterior urethral valves (PUV): only males 1-2% • Urethral agenesis (rare) • Ectopic ureter, ureterocoele (5-7%) <u>Non obstructive</u> • Vesicoureteral reflux (VUR) 10-12% • Nonrefluxing nonobstructive megaureter • Prune belly syndrome (rare) <u>Physiological:</u> • Physiological transient dilation of collecting system (41-88%): resolves in pregnancy	<u>**Syndromes**</u> • **Meckel Gruber**: cleft lip/palate, encephalocele, polydactyly and microphthalmia, heart defects and ambiguous genitalia, Autosomal recessive 17q21-q24 or 11q13. • **Beckwidth-Weidermann**: overgrowth syndrome, exomphalos, Autosomal dominant 11p15.5 • **Simpson-Golabi-Behmel:** overgrowth syndrome X-linked /mutations in GPC3 • **Bardet Biedl:** genital anomalies and polydactyly Autosomal recessive 2q, 3p,11q,15q and 16q • **Pearlman:** Diaphragmatic hernia, macrosomia, cleft palate, dextrocardia, cryptorchidism, Autosomal recessive • **Zellweger**: hepatomegaly, hypotonia, cataracts Autosomal recessive, multiple genes • **Elejalde**- nasal hypoplasia, craniosynostosis, exomphalos, short limbs, hydrops • **Di George**: cardiac anomalies, clefts, absent thymus, 22q deletion • **VACTERL:** vertebral anomalies, anal stenosis or atresia, tracheo-esophageal fistula, radial, renal, cardiac and non-radial limb defects. Sporadic <u>**Aneuploidies**</u> • Trisomy 8,9,13,18 and 21, 45X, Triploidy	• Normal Karyotype • Not associated with syndromes • Aetiology nonspecific
Antenatal management	• Genetics referral • Invasive testing should be offered • Ideally microarray and specifically request PKHD1 gene on chromosome 6P21 (80% cases) • Parenteral karyotyping may be indicated • Discuss prognosis and option for termination pregnancy in early onset disease • Serial ultrasounds to assess liquor volume • Renal and paediatric consultation	• Morphological fetal assessment for other anomalies and syndromes • Invasive testing is offered if bilateral renal involvement or if associated non-renal abnormality • Serial US to check liquor volume and contralateral kidney • Renal and paediatric consultation • Refer parents and siblings for renal ultrasound	• Genetic consultation • Can offer gene sequencing and identification of mutations with microarray (DR 50 to 75% for PKD1 and 75 % for APKD2) • If no family history, tuberous sclerosis should be excluded • Serial ultrasounds to assess liquor volume • Renal and paediatric input • Refer parents and siblings for renal ultrasound	• Degree of hydronephrosis is significant: AP diameter >4mm in 2nd and 7mm in 3rd trimester. Postnatal risks greater if >6mm in 2nd and >10 in 3rd trimester • Isolated mild pyelectasis doesn't incr risk of aneuploidy • Offer karyotype if other anomalies • Oligohydramnios in 2nd T: poor prognosis, can offer TOP • Serial US: monitor bladder distension and AFI • Can assess fetal urinary component in specialized centers to determine which fetuses may benefit from intervention: healthy fetus-hypotonic urine, progressive renal damage –isotonic urine • Fetal intervention (vesicoamniotic shunting and fetal cystoscopic ablation of PUV) improves lung function but not of renal benefit. • Renal and paediatric input	• Genetic counselling • Offer karyotype with microarray • TOP may be indicated • Serial ultrasounds to assess liquor volume	• Genetic analysis may be indicated • Serial ultrasounds to assess liquor and kidneys • Often transient
Outcomes	<u>Prenatal</u>: • Can develop renal failure in utero • 40–50% develop hepatic fibrosis. <u>Postnatal:</u> • 30% die in neonatal period • 55-100% develop hypertension by 15 years of age • 20-45% have end stage renal disease by 15 years of age • 11-45% develop portal hypertension • May need dialysis and renal transplantation	<u>Unilateral</u> • Usually managed conservatively • May undergo spontaneous regression and atrophy • Nephrectomy of affected kidney: last resort if recurrent infections, haematuria or severe HPT. • Involution of cysts occurs in 25% of cases in 1st 2 years. Antenatal involution of cysts rarely occurs. • Often recurrent UTI- need antibiotic prophylaxis • Must have postnatal follow up of the contralateral kidney <u>Bilateral</u> • Most develop chronic renal failure: dialysis and renal transplantation	• Recurrent urinary infections • Gross hematuria (40% adults) • Cystic hepatic and liver disease: relatively common in adults. • 50% will progress to ESRD needing dialysis and renal transplant <u>ADPKD1:</u> Hypertension in 1st year of life <u>ADPPKD2:</u> Asymptomatic until later in life Develop hypertension, renal failure, hemangiomas and aneurisms	• Morbidity and mortality directly linked to cause • Unilateral: survival 100%, may require surgery later in life • Bilateral with oligohydramnios: worst outcome, develop pulmonary hypoplasia and compression deformities of skeletal system • Oligohydramnios in the 2nd trimester usually fatal	• Dependent on associated condition	• No postnatal abnormalities • Excellent outcome • Possible small risk of ADPKD in adulthood
Reoccurrence risk	25%	Small (2-3%) unless associated with genetic syndrome	50% if one parent is affected	Very small	Dependent on associated condition	Unknown