

Disclaimer: These notes were taken by Ruth Dameron at the LMBBS Conference in Houston, June 17, 2006. There has been no attempt to verify accuracy. Do not quote the speakers based on these notes!

What are the kidney problems? Why are they there? What can be done to manage them?

(See notes from Dr. Nico's talk – he has essentially proven that the cilia genes cause the developing kidney cells to fail to differentiate – too many of one function, not enough (or none) performing another necessary function.)

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What do normal kidneys do?

What do we know about kidney problems of BBS

What is kidney failure, dialysis and transplant?

Every language has a different word for kidney.

English – kidney

Latin – ren (we call kidney disease *renal* diseases)

Greek – nephro (the study of kidneys is called *nephrology*)

The kidney is the master chemist of the body

- disposes of waste products of daily metabolism
- maintains water balance
- produces renin, hormone which regulates bp
- regulates body salts (Na, Cl, K)
- regulates acid-base balance
- regulates minerals (calcium, magnesium, phosphorous); interacts with glands and bones
- Produces active form of vitamin D from what is taken in in diet or skin
- Produces erythropoietin, a hormone that stimulates bone marrow to produce red blood cells and prevent anemia
- Removes drugs/medicines & toxins

Your kidney is the best sewage treatment plant ever designed!

Kidney disease can lead to:

- Bone disease or poor tooth development
- Rickets
- resistance to growth hormone
- edema (swelling) from mismanaging salts
- high blood pressure
- anemia
- dialysis or transplant ultimately

Kidney has a component called the nephron: filters blood; output is urine; leads to a tubule lined with different cells to do different things along the way. Finally collects the urine you want to get rid of and channels it to the ureter to the bladder.

Kidney problems in BBS:

Don't know the incidence. If you don't look very hard, probably 25%. If you screen everyone, it could be as high as 70 to 90%. But not everyone is looked at because not everyone has noticeable symptoms.

Mostly BBS causes structural abnormalities. It also causes some functional ones.

- Renal cysts – any fluid filled sac in the tubule or nephron; can be microscopic or large; many hereditary diseases have renal cysts.
- multicystic – multiple cysts of varying sizes – most common abnormality in BBS
- polycystic is a completely different disease, not found in BBS (Dr. Nico disagrees. “BBS can cause the same kind of cyst as what is in polycystic disease.”)
 - usually asymptomatic; can be confused with carcinoma; need CT scan
- Fetal lobulations – fetus starts out with lobulation when young, smoothes out over time; but in BBS, it doesn't smooth out.
- dysplasia, absent kidney, abnormally placed kidney – alternate ways in which the kidney is formed.
 - Doesn't grow to normal size; often found as a small kidney; can be one or both of them;
 - primitive filters
 - primitive tubules
 - cartilate and muscle tissues which don't even belong there
- cysts are frequent – not enough normal kidney to get proper function
- kidney stones
- bladder reflux (vesicoureteral reflux)
 - reflux means some urine goes UP when you void, not just down
 - some never gets out and causes urinary tract infections and causes scars on the kidney
 - diagnose with dye in bladder and watch which direction it goes
- ureteral duplication
- bladder problems needing catheters

Kidney problems in BBS – what we know

Somewhere between 25 to 70% have some kind of kidney issue – some are problematic; some are not.

infants, children, and adults

Sometimes they are seen in prenatal ultrasound, may look better by 2 years old but may not really be ok.

To find, often won't have certain symptoms,

- need x-ray or ultrasound
- 50-60% have high blood pressure
- 10-50% chronic renal failure CRF

If there is end stage kidney failure – it's time for dialysis or transplant

hemodialysis – done in a dialysis center

home peritoneal dialysis – done with a machine or manually with a special tube in the abdomen

transplant

How do you detect problems?

- Often no symptoms
- polyuria (urinating frequently or a lot of quantity), anemia
- kidney or bladder infections
- family history
- positive prenatal ultrasound
- real way to find it by screening
 - blood pressure annually
 - urinalysis every year annually
 - blood test for anemia and kidney function annually
 - ultrasound