

Outline

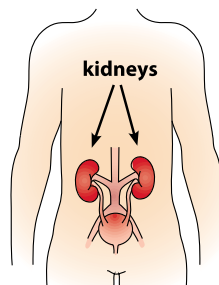
Kidney Symptoms and Medical Follow-up

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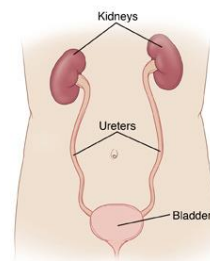
- Basics of kidney structure and function
- Kidney problems in patients with 17q12 deletion or duplication
- Testing for kidney disease
- Treatment and follow-up

Where are the kidneys?

- The kidneys are located under the lower ribs in the back of the body above the waist
- They are about the size of your fist
- They **make urine**



Parts of the Urinary Tract



What do the kidneys do?

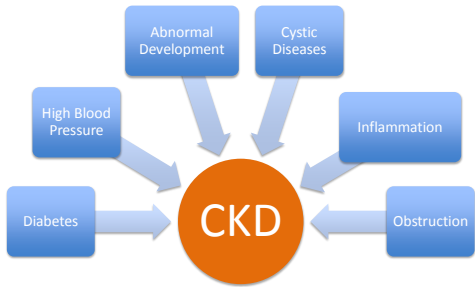
Three very important jobs!

- Filter or “clean” the blood
- Keep the right amount of fluids in your body
- Make hormones that help:
 - Make red blood cells
 - Control blood pressure
 - Keep bones strong

Kidney Disease

- If the kidneys get damaged they can't do all the things that they should
- This is called chronic kidney disease or CKD

What can damage the kidneys?



How are the kidneys affected in 17q12 deletions or duplications?

- Abnormal development - “congenital anomalies”, detected on ultrasound
 - Kidney cysts
 - Kidney hypoplasia-> small but normal kidney
 - Kidney dysplasia -> abnormal kidney tissue
 - Kidney agenesis -> born with only 1 kidney
- Reduced kidney function
 - Unclear timeline
 - Not everyone

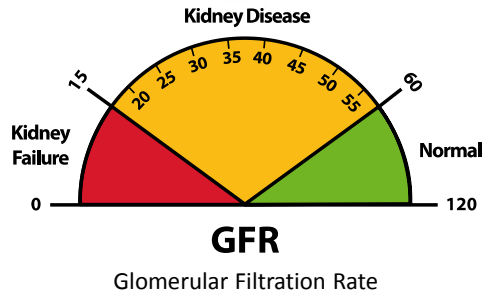
How do you test for kidney disease?

Three simple tests:

1. Blood test for “GFR”
2. Urine test for protein
3. Blood pressure

Early kidney disease often has no symptoms!

1. Blood test for GFR



2. Urine albumin (protein)



3. Blood Pressure



Complications of Kidney Disease

- Fluid build up
- High potassium levels
- Anemia
- Weak bones
- High blood pressure
- Malnutrition
- Growth restriction
- Cognitive disturbance

How is kidney disease treated?

- Blood pressure control
- Diabetes control
- Treatment of protein in the urine
- Avoidance of certain pain medications
- Dietary advice differs based on child/adult and stage of kidney disease

Early treatment can slow disease progression

Pain Medicines to Avoid

- Ibuprofen (Advil, Motrin)
- Naproxen (Aleve)
- Celebrex
- Toradol

Also note that certain medications may need to be dosed differently if you have kidney disease

What happens if the kidneys fail completely?

- Kidney failure is called End Stage Renal Disease or ESRD
- Options include:
 - Hemodialysis
 - Peritoneal dialysis
 - Transplant -> best survival

Remember

- A variety of kidney malformations can occur in patients with 17q12 deletion or duplication
- Not everyone develops kidney disease
- 3 simple test can be used to screen for kidney disease
- Early detection can slow disease progression and prevent development of ESRD

Resources

- National Kidney Foundation www.kidney.org
- National Kidney Disease Education Program nkdep.nih.gov
- American Kidney Fund www.kidneyfund.org

Questions?