Kidney Symptoms and Medical Follow-up

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Outline

• 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?

- Abnormal Development
- Cystic Diseases
- High Blood Pressure
- Inflammation
- Diabetes
- Obstruction

CKD

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

   Glomerular Filtration Rate

   - Kidney Failure
   - Normal
   - 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](http://www.kidney.org)
- American Kidney Fund [www.kidneyfund.org](http://www.kidneyfund.org)
Questions?