

## Outline

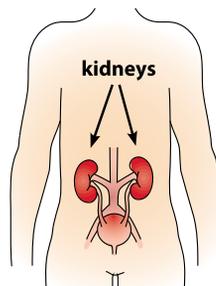
### Kidney Symptoms and Medical Follow-up

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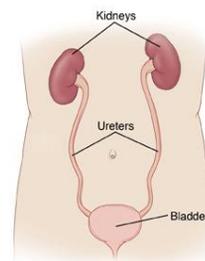
- Basics of kidney structure and function
- Kidney problems in patients with 17q12 deletion or duplication
- Testing for kidney disease
- Treatment and follow-up

### 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?

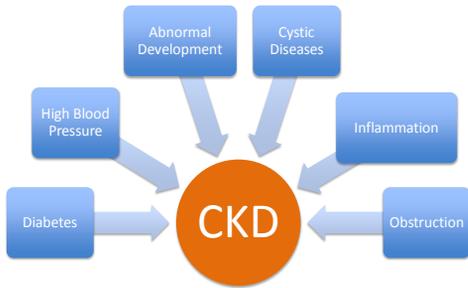
Three very important jobs!

- 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

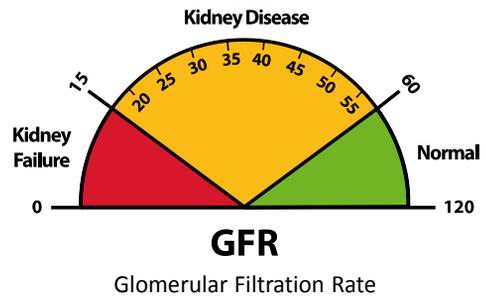
## How do you test for kidney disease?

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](http://www.kidney.org)
- National Kidney Disease Education Program [nkdep.nih.gov](http://nkdep.nih.gov)
- American Kidney Fund [www.kidneyfund.org](http://www.kidneyfund.org)

Questions?