Pocket Guide to Thinking About Genetics and Kidney Disease

A tool to help patients and care partners have more effective discussions with their healthcare team about possible underlying genetic causes of their kidney disease.

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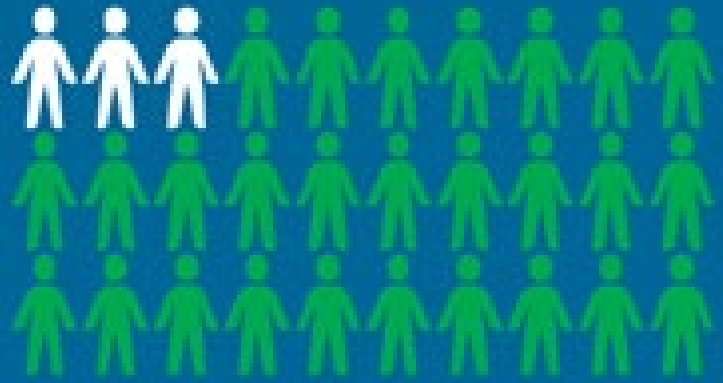
Did you know that up to **10% of all chronic kidney disease (CKD) patients** in the U.S. – as many as **3.7 million people** – have a genetic mutation that causes their disease?



This staggering number includes an estimated **40% of dialysis patients** who, along with their nephrologists, are in the dark about the cause of their kidney failure.

If CKD runs in your family, you may have an inherited kidney disease.

What is next if I suspect my kidney disease may be caused by a genetic condition?

Inherited kidney diseases (IKDs) are among the leading causes of early-onset chronic kidney disease (CKD) and are responsible for at least 10–15% of cases of kidney replacement therapy (KRT) in adults[1].

- If you think you might have an increased risk for an underlying genetic cause for your kidney disease, talk to your nephrologist (kidney doctor) or another key healthcare team member.
- Your nephrologist can review your medical and family history with you to evaluate your individual risk of having an underlying genetic condition.

- If they feel that genetics could be involved in your kidney disease, they can discuss genetic testing options with you.
- After your discussion, your nephrologist will either order genetic testing or refer you to a genetic counselor or geneticist for counseling and testing.

[1]*Roser Torra, Mónica Furlano, Alberto Ortiz, Elisabet Ars, Genetic kidney diseases as an underrecognized cause of chronic kidney disease: the key role of international registry reports, Clinical Kidney Journal, Volume 14, Issue 8, August 2021, Pages 1879–1885, https://doi.org/10.1093/ckj/sfab056.*

What if your nephrologist does not feel you have an underlying genetic cause for your kidney disease?

Ask them why they do not feel you have an underlying genetic cause. If the answer makes sense, then consider reevaluating in the future with them. If the answer does not make sense, or you still have concerns, you have every right to seek a second opinion.

Questions to ask your nephrologist before you have genetic testing:

1. Do you plan to order testing for just one genetic condition or many on a panel?
2. How will the genetic testing be conducted? Blood test? Salvia test?
3. How long will it take to get results?

4. Is there a chance the testing will be inconclusive without a specific answer? If yes, what will be done next?
5. Does my insurance cover genetic testing? If so, what is my out-of-pocket cost? If not, what is the cost?
6. How will you tell me the results?
7. Who will have access to my results?
8. Is there a genetic counselor who can talk with me about my results?
9. Should I get life insurance before having genetic testing?
10. Who else in my family might have a genetic condition if I do?
11. What will genetic testing tell me about my kidney disease?
12. How will you change my care if I have a genetic cause for my kidney disease?

Scan QR Code to see some common underlying genetic types and causes of kidney disease?

What are some ways in which genetic causes for kidney diseases can run in families?

Autosomal dominant - A pattern of inheritance for genetic traits or conditions in which a disease-causing gene change from one parent is enough to cause the symptoms of the condition. Typically, there is someone with this condition in every generation.

Autosomal recessive - A pattern of inheritance for genetic traits or conditions in which two disease-causing gene changes in the same gene (one from each

parent) is required to cause the symptoms of the condition. Often conditions that are autosomal recessive seem to be diagnosed without anyone else in the family having the condition. The siblings of the affected person are at highest risk of having the condition too.

X-linked - A pattern of inheritance for genetic traits or conditions in which a disease-causing gene change is located on the X-chromosome. Most often a disease-causing gene change on the X-chromosome from one parent is enough to cause the symptoms of the condition. Typically, there is someone with the condition in every generation although men may come to medical attention first with more severe symptoms.

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