The Importance of Family History

"I'd like to ask a few questions about the health of your family because knowing your relatives' medical histories will help us provide you with the most appropriate care."

This simple invitation should help patients understand the importance of a detailed family history. Explaining the potential benefits and implications of making a family pedigree may assist the patient in making an informed decision about their care. For example, patients may harbor misconceptions about how diseases are inherited, the risk of inheriting a condition or illness, and the implications for family planning.

In the current day of modern genomics, a clinician does not need to be an expert in genetics to consider genetic causes of a disease and assess the risk of a hereditary disease. A careful family history is part of a routine medical visit and should take no more than a few minutes. In Mendelian genetic disorders, it is one of the most cost-effective screening tests, and can yield information not otherwise apparent or volunteered by patients. A simple pedigree may help with a diagnosis, deciding on laboratory tests, calculating risks for inheritance, informing reproductive options, and making decisions about surveillance or preventive measures. Pedigrees may be helpful in identifying other genetic causes of renal cysts from recessive polycystic kidney disease (PKD) or X-linked disorders such as Fabry disease.

The history usually needs to be collected once for an entire family and provides an important living document and component of the medical chart. Obtaining an accurate and detailed family history is indispensable in the care of families with PKD.

What to Do

Detailed information should be gathered from the patient about first-degree relatives (parents, siblings, and offspring), including their age or date of birth, vital status, date and age at death, cause of death, and any known diseases. Critical to PKD, note those at-risk family members who have undergone screening, the type of imaging performed, and the age the screening took place. As with all pedigrees, it is important to identify full vs half siblings, adopted children, pregnancy outcomes, and monozygotic vs dizygotic twins. Use of the attached questionnaire may be helpful to gather basic information from patients prior to the interview.

Evaluate the significance of any illnesses among family members and whether they are genetic in nature. With regard to PKD, this would include date of diagnosis of ADPKD, end-stage renal disease, and kidney transplant if indicated, whether at risk persons have been screened, and the imaging method used.

Extend the inquiry to additional generations. It is important to identify consanguineous relationships. Delicate ways to address this issue include asking "Do you and your partner have any of the same relatives?" or "Is anyone in your family related to anyone in your partner's family, other than by marriage?" Asking about geographic location of families and surnames may also shed light on consanguinity.

Prepare a pedigree using the template provided. Complete a row for each generation, indicating the informant with an arrow. Indicate by shading the persons in the family who have had or were suspected to have a disease or disorder. Take note of appropriate symbols for twins, consanguinity, and stillbirths. Attempt to identify paternity outside marriage, which can cause confusion in interpretation if omitted. It may take several visits before outside paternity can be established for a patient with no antecedent family history of the disease. The one who knows for sure is the mother of the newly discovered offspring. The more family members that are included, the greater the predictive value of the pedigree.

Typical pedigrees for PKD are shown in the Figure. In autosomal dominant PKD (top), affected males and females have an equal probability of passing the trait, and each offspring has a 50% chance of inheriting the mutant allele. In autosomal recessive PKD (bottom), the disease does not develop in carriers with a single mutant allele. Each child born from two carrier parents has a 25% chance of inheriting two mutant alleles and being affected.

Additional Resources


March of Dimes: Genetics and Your Practice. www.marchofdimes.com/gyponline. Provides resources and tools, information on testing and screening, family health, and referrals.


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Supported by an educational grant from Otsuka America Pharmaceutical, Inc. ©2013 CMEology

Disclaimer: This article is for information only and should not be used for the diagnosis or treatment of medical conditions. This information is not a substitute for genetic counseling or as a guideline for patient management.
## Family History Questionnaire

**Name:**

**Date:** ___________________  **Date of Birth:** ___________________

### Biologic Father

<table>
<thead>
<tr>
<th>Name</th>
<th>Yes Living?</th>
<th>Current age or birth date (at death)</th>
<th>Health</th>
<th>Cause of death or known illness at time of death</th>
</tr>
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<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>☐ Good</td>
<td>☐ Fair ☐ Poor</td>
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</tbody>
</table>

- **Health problems**

- **If he/she had ADPKD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________
- **If he/she had ESRD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________

### Biologic Mother

<table>
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<tr>
<th>Name</th>
<th>Yes Living?</th>
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- **If he/she had ADPKD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________
- **If he/she had ESRD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________

### Mother's Side of Family

#### Grandmother

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  - **Imaging test:** ____________  **Date of diagnosis:** ____________

### Father's Side of Family

#### Grandmother

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- **If he/she had ADPKD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________
- **If he/she had ESRD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________

### Aunts and Uncles

#### Aunts

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- **If he/she had ADPKD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________
- **If he/she had ESRD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________

#### Uncles

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- **Health problems**

- **If he/she had ADPKD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________
- **If he/she had ESRD:**
  - **Imaging test:** ____________  **Date of diagnosis:** ____________

### Abbreviations:

- ADPKD = Autosomal Dominant Polycystic Kidney Disease
- ESRD = End-Stage Renal Disease (Kidney Failure)

Instructions:

Please provide information on only biologic relatives; do not include step-relatives, adopted, or foster parents. Indicate half-brothers and half-sisters with an asterisk (*). If exact ages are not known, use approximate age range. Use additional pages as needed.
### Brothers

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</table>

If he/she had ADPKD: Imaging test: Date of diagnosis:

### Sisters

| Name | Current age or birth date | Health | Living? | Age (date) at death | Cause of death or known illnesses at time of death | Health problems |
|------|---------------------------|--------|---------|---------------------|================================================|----------------|
|      |                           |        |         |                     |                                                  |                |

If he/she had ADPKD: Imaging test: Date of diagnosis:

### Sons

| Name | Current age or birth date | Health | Living? | Age (date) at death | Cause of death or known illnesses at time of death | Health problems |
|------|---------------------------|--------|---------|---------------------|================================================|----------------|
|      |                           |        |         |                     |                                                  |                |

If he/she had ADPKD: Imaging test: Date of diagnosis:

### Daughters

| Name | Current age or birth date | Health | Living? | Age (date) at death | Cause of death or known illnesses at time of death | Health problems |
|------|---------------------------|--------|---------|---------------------|================================================|----------------|
|      |                           |        |         |                     |                                                  |                |

If he/she had ADPKD: Imaging test: Date of diagnosis: