

Genetic Testing and Referral Criteria

for Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Based on existing data, genetic testing is not always necessary for diagnosis or selecting treatment options for patients with a clinical diagnosis of ADPKD. There are specific situations, however, where genetic testing may be helpful. There are two ways that genetic testing can be accessed in BC; the nephrology team can order the testing directly, or it can be done through involvement and referral to medical genetics.

The main consideration when requesting publicly funded genetic testing is that it should only be done in situations where that testing will **change patient management**; in those settings genetic testing is desirable and encouraged but the criteria outlined in Table 1 refers to situations where the testing may not change management and therefore would not be recommended.

The following are situations in which genetic testing is **not** typically pursued by BC Nephrologists as recommended by the BC Renal Medical Advisory Group:

- 1. For diagnosis of ADPKD where imaging and/or family history provides sufficient information to diagnose ADPKD without the need for genetic testing**

For more details, see [Best Practices: Care of Patients with Autosomal Dominant Polycystic Kidney Disease in BC's Kidney Care Clinics](#)

- 2. Diagnosis is not 100% confirmed, but the results of genetic testing would not change management**

- For example, a patient with an uncertain diagnosis, but predicted to have an indolent disease course irrespective such that testing would not alter clinical management; eg, an older adult with well-preserved kidney function and minimally enlarged kidneys who would not be a candidate for targeted ADPKD treatment regardless of genetic results
- Another example may be in asymptomatic children, teens and young adults from a family with known ADPKD and a negative ultrasound which does not completely rule out the possibility of ADPKD; in this case some may consider genetic testing but an alternate acceptable strategy may be to repeat imaging later in life.

Table 1 outlines when publicly funded genetic testing may be considered, information required and recommendations.

Table 1

Considerations for Genetic Care in ADPKD	Recommendations	Information Required
<p>1. ADPKD is the most likely consideration but no family history and morphology are not sufficiently typical/characteristic to diagnose ADPKD</p> <p style="text-align: center;"><u>AND</u></p> <p>Clarity around the diagnosis is needed for management/ treatment decisions such as candidacy for disease modifying treatment</p>	<ul style="list-style-type: none"> If nephrology team is comfortable, genetic testing can be ordered directly* Before ordering, ensure genetic testing has not already been ordered or is not currently in progress Apply for out of province/ country PKD Panel (pre-filled link) Provide BC Renal's "Screening and Testing for ADPKD" resource to patient 	<p>Standardized requisition package:</p> <ul style="list-style-type: none"> Consent for Release of Information Out of Province/Country Lab and Genetic Testing Services Funding Application Pre-filled for ADPKD BC Renal's Screening and Testing for ADPKD <p>Medical Genetics Referral form:</p>
<p>2. Unusual radiographic appearance where other cystic renal diseases are possible or likely (eg, TSC, ARPKD, etc)</p> <p style="text-align: center;"><u>AND</u></p> <p>Clarity around the diagnosis is needed for management/ treatment decisions such as candidacy for disease modifying treatment</p>	<ul style="list-style-type: none"> Medical genetics referral Medical genetics consideration of PKD Panel or Cystic Kidney Disease Panel 	<ul style="list-style-type: none"> For Island Health Medical Genetics For Provincial Medical Genetics Program at B.C. Women's Hospital
<p>3. Family planning, especially where it may influence the decision to conceive or pursue pre-implantation diagnosis</p>	<ul style="list-style-type: none"> Medical genetic assessment and counselling 	
<p>4. Work-up of family members being considered as kidney donors where greater diagnostic certainty is required for approval of a donor</p>	<ul style="list-style-type: none"> Medical genetics referral arranged by donor team Typically known variant testing only 	
<p>5. Other situations that do not fall under consideration #1</p>	<ul style="list-style-type: none"> Case by Case 	

*In consideration #1 it is reasonable for the nephrology team to order the testing directly if they are comfortable with the testing itself as well as counselling the patient and family about the implications of this prior to testing. For other considerations or any situation where the nephrology team is uncertain about the implications of testing or would like further input, a medical genetics referral is advised. For more information about genetic testing in BC including how to order, please visit the Division of Genome Diagnostics website under policies and protocols: genebc.ca and the Provincial Medical Genetics program bcwomens.ca/our-services/medical-genetics

For more information about genetic testing in BC including how to order, please visit the **Division of Genome Diagnostics** website under policies and protocols: genebc.ca and the **Provincial Medical Genetics program** bcwomens.ca/our-services/medical-genetics