Ordering Guidance: Genetic Testing for Chronic Kidney Disease

1. Confirm Patient Eligibility

For detailed eligibility criteria, see **Chronic Kidney Disease Genetic Testing Recommendations**.

2. Select Appropriate Genetic Test (see also Figure 1)

When choosing the most appropriate test for a patient and/or family, the test should be sufficiently comprehensive to encompass the genes associated with the differential diagnoses under consideration. While standardized criteria support consistency and equity, test selection and the decision to test should be guided by clinician's expertise, diagnostic certainty and understanding of the individual's presentation, and family history.

3. Obtain Consent and Complete Ordering Documents

Complete laboratory requisition for one of the laboratories below.

Labs Offering Kidney Genetic Testing	Link to Laboratory Requisition
Mount Sinai Hospital	Link
University Health Network	Link

4. Arrange Sample Collection

a) Patients can have bloodwork done at a community lab or a hospital site. To find a community lab site, visit Ontario Association of Medical Laboratories. The lab will transport the sample to the testing laboratory.
b) The laboratory requisition includes instructions on sample type and sample collection information.
c) Give your patient the information sheet for Genetic Testing for Chronic Kidney Disease.

5. Receive results and disclose/follow-up accordingly

For further assistance in test selection, ordering, result interpretation, high suspicion of a genetic diagnosis after uninformative results, and management recommendations, please consider the following options as needed:

- For patient-specific medical advice:
  - Submit an eConsult request to connect with a genetics specialist through OTNhub
  - Send a referral to your local genetics clinic (Ontario Genetics Clinic Directory)
- For test navigation support, education and resources:
  - Contact gcConnect (1-844-564-4363, gcConnect@ontariohealth.ca)

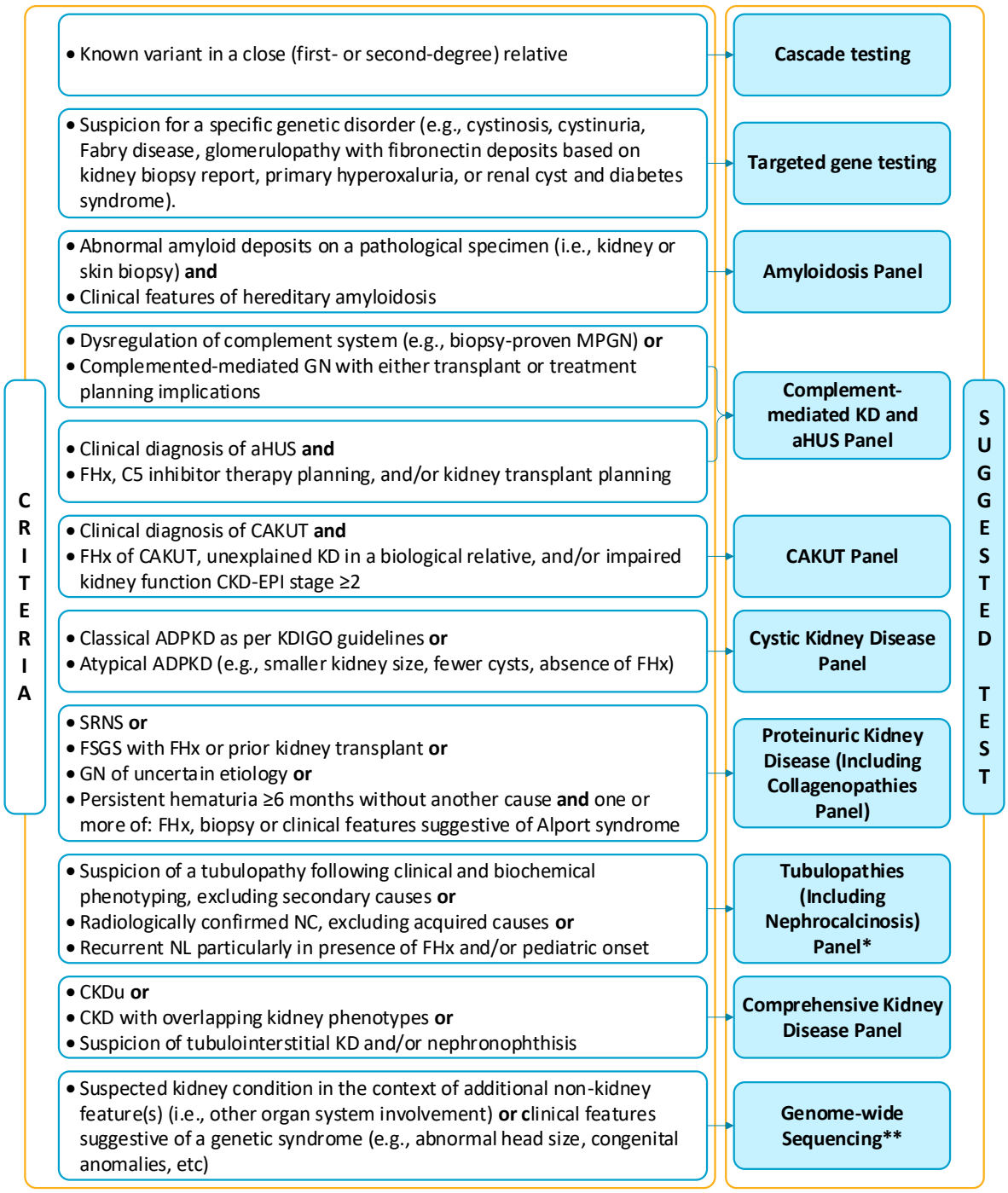

Figure 1. Testing Options for Suspected Subtypes of CKD

* Consider a sub-panel within the tubulopathies panel if there is a high index of suspicion for a specific phenotype. ** Although *MUC1* is included on the comprehensive kidney disease panel, the most common pathogenic variants are not detectable using standard sequencing methods. Seek input from a kidney genetics expert regarding *MUC1* testing (e.g., via eConsult or referral to a local genetics clinic). **ADPKD**, autosomal dominant polycystic kidney disease; **aHUS**, atypical hemolytic uremic syndrome; **CAKUT**, congenital anomalies of the kidney and urinary tract; **CKD**, chronic kidney disease; **CKD-EPI**, Chronic Kidney Disease Epidemiology Collaboration; **CKDu**, chronic kidney disease of unknown etiology; **FHx**, family history; **FSGS**, focal segmental glomerulosclerosis; **GN**, glomerulonephritis; **KD**, kidney disease; **MPGN**, membraneproliferative glomerulonephritis; **NC**, nephrocalcinosis; **NL**, nephrolithiasis; **SRNS**, steroid resistant nephrotic syndrome. Please note *APOL1* genotype is included in both the Proteinuric Kidney Disease (including Collagenopathies) Panel and Comprehensive Kidney Disease Panel. For further details, please see the link for more information: ontariohealth.ca/clinical/genetics/guidance.