Congenital Anomalies of the Kidneys and Urinary Tract (CAKUT)

THE GENOMIC INSTITUTE

Step 1

Document the following clinical information to assist in phenotyping:

- Kidney function
- Kidney imaging
- Extrarenal features
- Family history of kidney disease, including kidney phenotype

For patients with a negative or unknown family history:
If possible, assess the phenotype of both parents with kidney imaging and kidney function testing.

For patients with a positive family history:
If a family member has had genetic testing and a disease-causing variant has been identified:

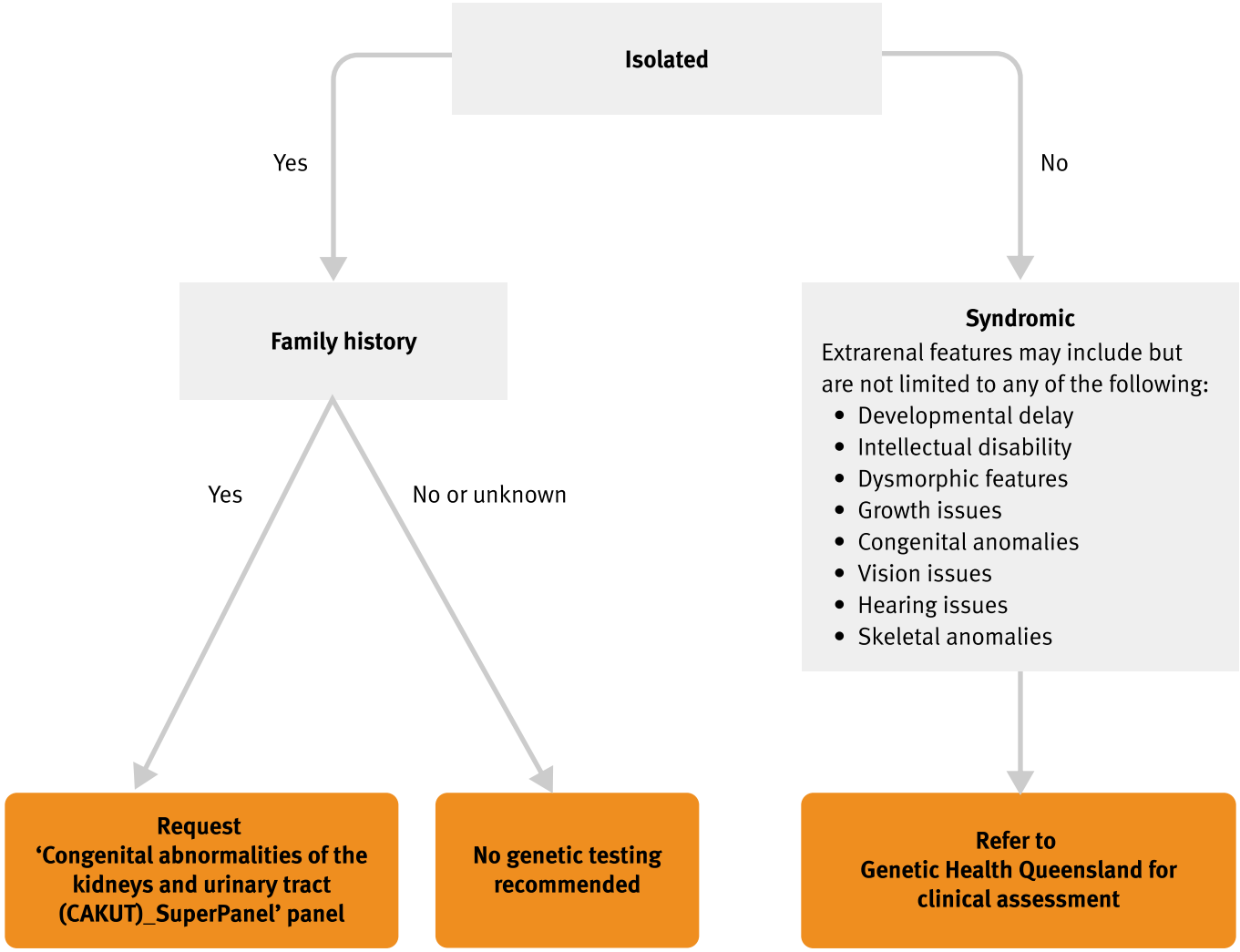
- Do not proceed to genomic testing
- Refer to Genetic Health Queensland to discuss targeted confirmatory genetic testing

Consider genomic testing if there are any of the following indications for testing:

- Diagnostic uncertainty
- Genotype-specific management
- Family planning
- Risk clarification for family
- Transplant planning (particularly if donor is a blood relative)

Step 2

The following pathway guides genomic testing based on family history and extrarenal features.



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For further specialist advice, email Renal Genetics MDT at Renal_Genetics_MDT@health.qld.gov.au