

## GENETIC TESTING FOR PRIMARY KIDNEY DISEASE

### Executive Summary

**Genetic questions regarding primary kidney disease diagnosis will be added to patient registration in ANZDATA. These questions will identify if testing has taken place, if this testing influenced the primary kidney disease diagnosis, and what the causative gene was, if known.**

The ANZDATA Advisory Committee and the Special Interest Group - Genetics, agreed to this new data collection element and are to implement this within the Online Electronic Data Collection tool (<https://services.anzdata.org.au>) and the A3 Survey Paper Form from Survey 74 - 2023.

### Background

Primary Kidney Disease (PKD) and whether a kidney biopsy was performed as part of the diagnosis process, is collected and reported at the time of entry to the Registry. The addition of three new variables regarding genetic testing provides an opportunity to identify if genetic factors have influenced the diagnosis of PKD. Responses to these questions would assist in identifying the frequency of genetic testing; if the test gave a diagnosis for the underlying kidney disease and which gene/CNV was identified.

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### Data Element

The new genetic questions are:

- Was there a genetic test performed for investigation of the patient's underlying primary kidney disease? Yes/No (Variable Label - Genetic Test)
- Did the test give you a diagnosis of the underlying primary kidney disease? Yes/No/Unknown (Variable Label - PKD Diagnosis from Test)
- What was the Causative Gene? Drop down list using Kindeyome Super Panel from the *Green List* - (Variable Label - Causative Gene)

These questions will be added to 'Patient Details' and will be available for all new patient registrations from Survey 74 (2023).

### Collection of Data Element

Data can be submitted via the usual data collection methods (paper form, online data entry, file import, or API technology).

Data collection is to be reported at registration of a new patient to ANZDATA, in real-time. The data fields will remain available for all open survey periods, results may be delayed in reporting from the laboratory. Subsequently the Primary Kidney Disease field will also be available for changes, if required.

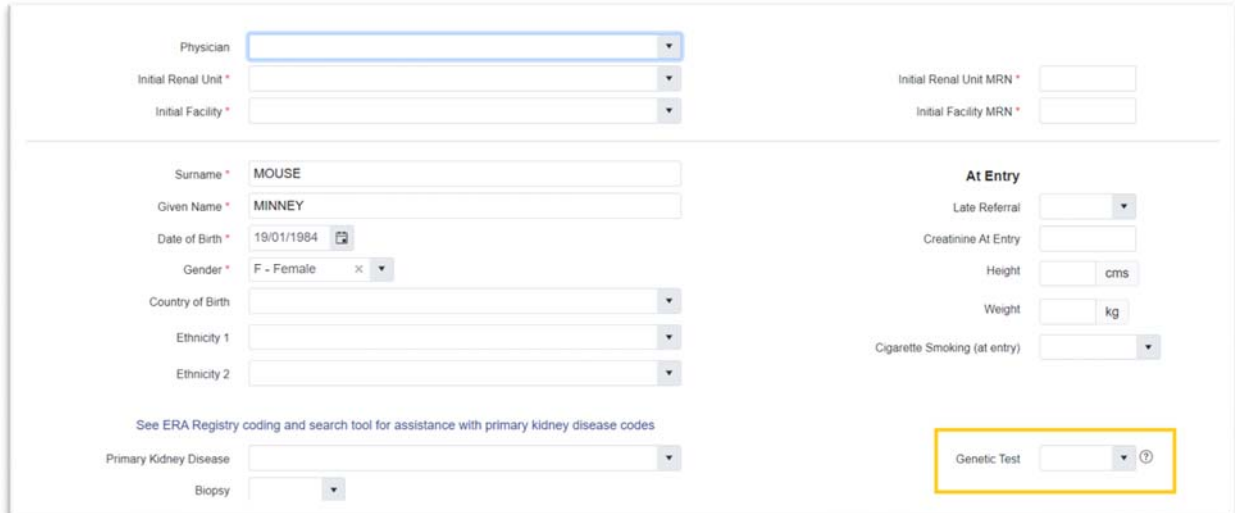
If data submission occurs by personnel other than a Physician, a Nephrologist may need to be consulted to confirm data integrity.

The data is required for collection from Survey 74 2023 for new patient registration.

# What's New in ANZDATA Data Collection

More technical specifications can be located from the ANZDATA Registry website:  
<https://services.anzdata.org.au/webservices>

**Figure 1 – Screen View – Patient Details – Genetic Test Question 1**



Physician

Initial Renal Unit \*

Initial Facility \*

Initial Renal Unit MRN \*

Initial Facility MRN \*

Surname \* MOUSE

Given Name \* MINNEY

Date of Birth \* 19/01/1984

Gender \* F - Female

Country of Birth

Ethnicity 1

Ethnicity 2

At Entry

Late Referral

Creatinine At Entry

Height  cms

Weight  kg

Cigarette Smoking (at entry)

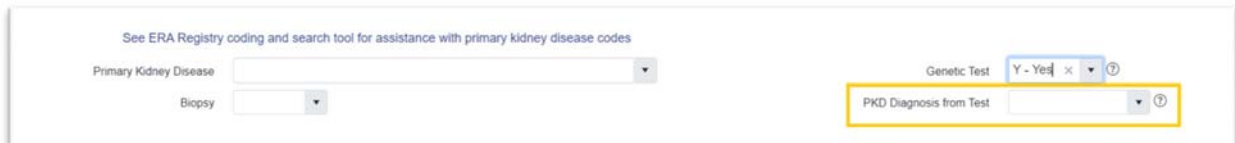
See ERA Registry coding and search tool for assistance with primary kidney disease codes

Primary Kidney Disease

Biopsy

Genetic Test

**Figure 2 – Screen View – Patient Details – Genetic Test Question 2**



See ERA Registry coding and search tool for assistance with primary kidney disease codes

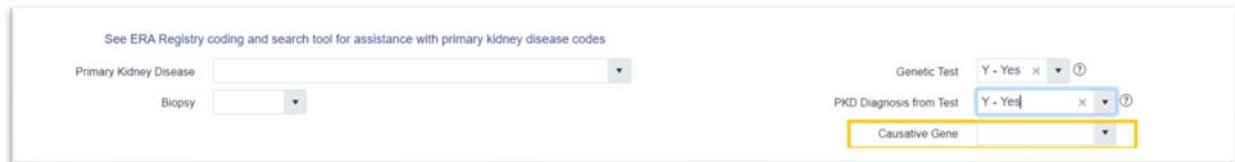
Primary Kidney Disease

Biopsy

Genetic Test Y - Yes

PKD Diagnosis from Test

**Figure 3 – Screen View – Patient Details – Genetic Test Question 3**



See ERA Registry coding and search tool for assistance with primary kidney disease codes

Primary Kidney Disease

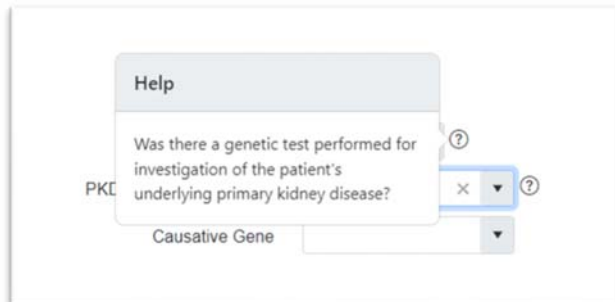
Biopsy

Genetic Test Y - Yes

PKD Diagnosis from Test Y - Yes

Causative Gene

**Figure 4 – Screen View – Patient Details – Genetic Test Question Help Text**



Help

Was there a genetic test performed for investigation of the patient's underlying primary kidney disease?

PKD Diagnosis from Test

Causative Gene