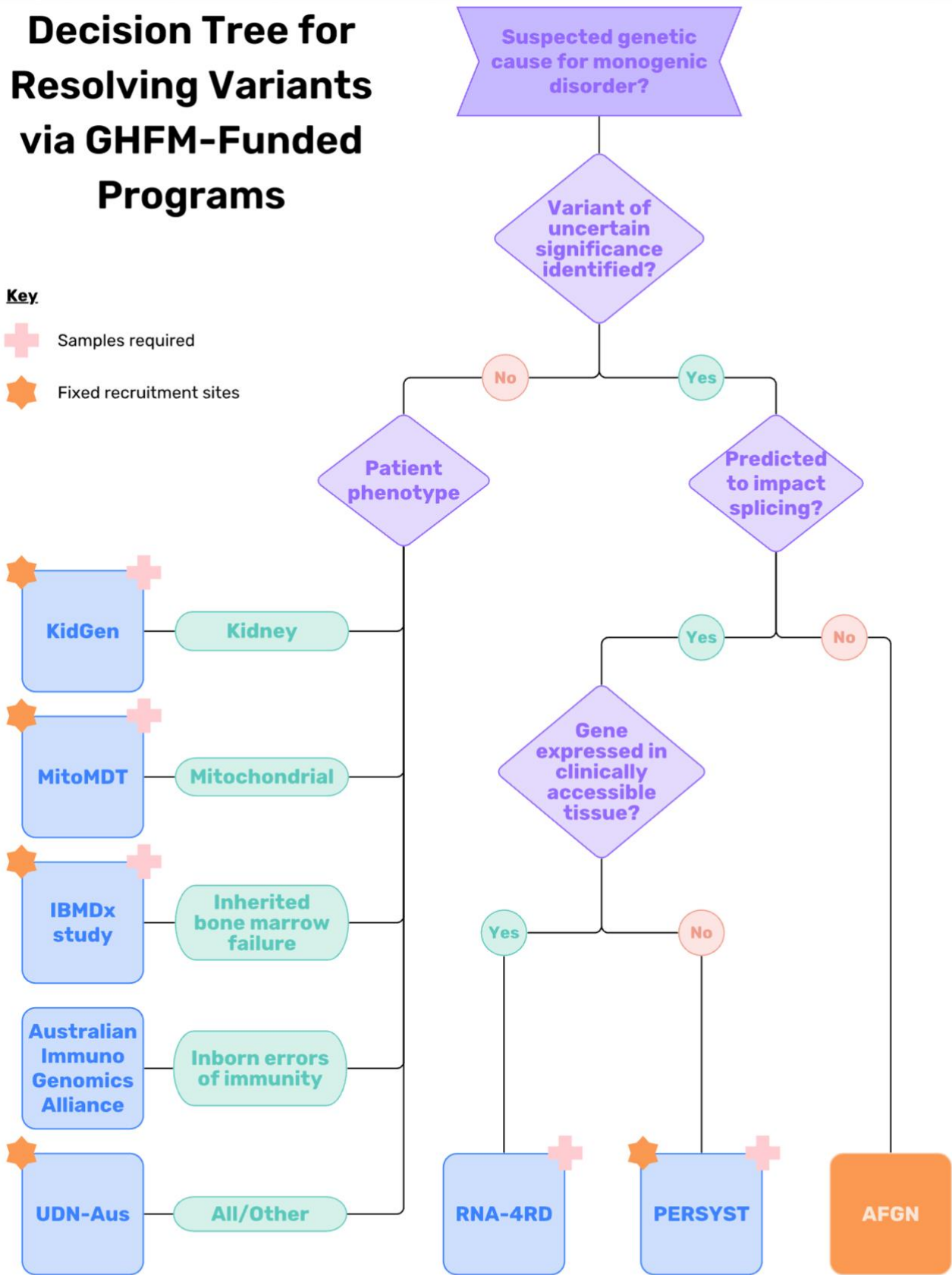


# Decision Tree for Resolving Variants via GHFM-Funded Programs

**Key**

- + Samples required
- ★ Fixed recruitment sites



★ +  
**RDMass Spec**  
 Referrals from VCGS, RDNOW, Acute Care & UDN-Aus only.

★  
**Ion channelopathy assays**  
 Patch-clamp assays for select ion channel genes.

★  
**MaveDB**  
 Functional assay database + workforce education.

# Program List

## **KidGen – National Kidney Genomics Program**

**PI:** Prof Andrew Mallett

**Contact:** Trudie Harris at [kidgen@mcri.edu.au](mailto:kidgen@mcri.edu.au)

**Recruitment:** July 2022 – Dec 2025

**More info:** [Visit AG webpage](#)

## **MitoMDT – Mitochondrial Diagnostic Network for Genomics and Omics**

**PI:** Prof David Thorburn

**Contact:** [Amanda.samarasinghe@mcri.edu.au](mailto:Amanda.samarasinghe@mcri.edu.au)

**Recruitment:** Until May 2025

**More info:** [Visit AG webpage](#)

## **IBMDx – Inherited bone marrow failure and related disorders study**

**PI:** Assoc Prof Piers Blombery

**Contact:** [Kelsey.Man@petermac.org](mailto:Kelsey.Man@petermac.org)

**Recruitment:** Feb 2022 – Dec 2024

**More info:** [Visit AG webpage](#)

## **Australian ImmunoGenomics Alliance (AIGA)**

**PI:** Prof Stuart Tangye

**Contact:** [s.tangye@garvan.org.au](mailto:s.tangye@garvan.org.au)

**Recruitment:** TBC

## **UDN-Aus – The Australian Undiagnosed Diseases Network**

**PI:** Prof John Christodoulou

**Contact:** Madeleine Harris at [udn.aus@mcri.edu.au](mailto:udn.aus@mcri.edu.au)

**Recruitment:** Until Oct 2024

**More info:** [Visit AG webpage](#)

## **RNA-4RD – Disease-agnostic, nationally-accessible pipelines of clinical RNA Diagnostics**

**PI:** Prof Sandra Cooper

**Contact:** [clynch-sutherland@cmri.org.au](mailto:clynch-sutherland@cmri.org.au)

**Recruitment:** Oct 2022 – Sept 2024

**More info:** [Visit AG webpage](#)

## **PERSYST – Pathogenic Evaluation of Recalcitrant Variants by Systematic Transactivation**

**PI:** Prof Josef Gecz  
& Dr Lachlan Jolly

**Contact:** [persyst@adelaide.edu.au](mailto:persyst@adelaide.edu.au)

**Recruitment:** Oct 2023 – Oct 2027

**More info:** [Visit AG webpage](#)

## **AFGN – Australian Functional Genomics Network**

**PI:** Prof Andrew Sinclair & Prof Sally Dunwoodie

**Contact:** [functional.genomics@mcri.edu.au](mailto:functional.genomics@mcri.edu.au)

**Recruitment:** Mar 2022 – Mar 2025

**More info:** [Visit AFGN website](#)

## **RDMassSpec**

**PI:** Assoc Prof David Stroud

**Contact:** [rosie.brown@mcri.edu.au](mailto:rosie.brown@mcri.edu.au)

**More info:** [Visit AG webpage](#)

## **High throughput functional genomics assays for ion channelopathies**

**PI:** Prof Jamie Vandenberg

**Contact:** [c.ng@victorchang.edu.au](mailto:c.ng@victorchang.edu.au)

## **Patch-clamp assays planned (2022-2026):**

SCN1A/2A/3A/4A/8A, KCNQ2/Q3, CACNA1A/1G/1H, KCNT1, CFTR, PKD1/2, KCNH2, SCN5A, KCNQ1

## **MaveDB**

**PI:** Dr Alan Rubin

**Contact:** [rehan.Villani@qimrberghofer.edu.au](mailto:rehan.Villani@qimrberghofer.edu.au)

**Remit:** Database for multiplexed assays of variant effect (MAVE) functional data.

Workforce education opportunities around functional evidence application in variant curation.