For paediatricians

Order Medicare-funded exome sequencing online

Online test request system

NOTE: Approval by clinical geneticists can be provided via this portal.

1 *Practitioner details*

- Fill out practitioner details.

2 *Patient details*

- For **eligibility criteria** see MBS Item Numbers 73358 & 73359.

  Select status of patient as **private**.

  **Please note: MBS funding is unavailable to patients seen in publicly (ie. state) funded clinics. If unsure regarding your patient's status, please clarify with your practice manager or head of department.*

- Select **sample** types:
  - saliva kits can be sent to family
  - choose 'DNA stored at VCGS' if previously tested at VCGS

3 *Test details*

- select: ***'Medicare testing'***

↳ ***'Childhood syndromes - diagnostic exome'***

- Acknowledge inclusion criteria:
  - Non-informative microarray complete
  - Satisfies clinical criteria

- Family type:
  Consider **TRIO** testing over singleton for a faster definitive result.
  - 8-10 week turnaround time
  - Shows parental inheritance

  **Note: both parents' names, DOB, and consent is required*

- Clinical features:
  Clinical notes - **detailed** clinical information is required for **accurate analysis**.

  Specify **severity** of ID/Global DD (if relevant) and microarray result.

  Include **history** of any affected relatives.

  Gene panels – *ID syndromic and non-syndromic panel* and *Mendeliome* are usually appropriate.

  - Consider additional panels for specific phenotypes
    (e.g. the *Macrocephaly Megalencephaly panel*)

Submit

You will be contacted to proceed with consent and sample collection.

Exome sequencing

Please contact our team for assistance.

E VCGSgenomics@vcgs.org.au
P 03 9340 9102