Head And Neck Paraganglioma: A Survey Amongst New Zealand Specialists

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Background

- Head and neck paraganglioma (HNPGL) with strong hereditary background (~40%)
- Succinate dehydrogenase genes (*SDHx*), mainly *SDHD* and *SDHB*, are the most common cause
- SDHB-deficient HNPGL more often metastasise than PGL carrying other driver genes
- International studies recommend early surgical treatment for *SDHx*-related HNPGL

Data gap in Aotearoa

- *SDHB* founder mutation R46Q suspected in Māori whanau
- No information in Aotearoa about
  - Frequency of hereditary HNPGL
  - Mutations
  - Clinical presentation
  - Clinical pathway / treatment

Study aims

1. Estimate the extent of referrals for genetic testing in patients with HNPGL
2. Engage with clinicians to better understand clinical pathways of patients with HNPGL in Aotearoa

Methods

- Anonymous survey for clinicians likely to encounter patients with HNPGL
- Survey distribution via email link to vocationally registered specialists listed with the New Zealand Medical Council in three specialties: otolaryngology (ORL), vascular surgery (VAS), and endocrinology (ENDO)
- Results were recorded in REDCap
- Incomplete entries (n=2) or obvious duplications (n=1) were removed before analysis (total number of entries 80)

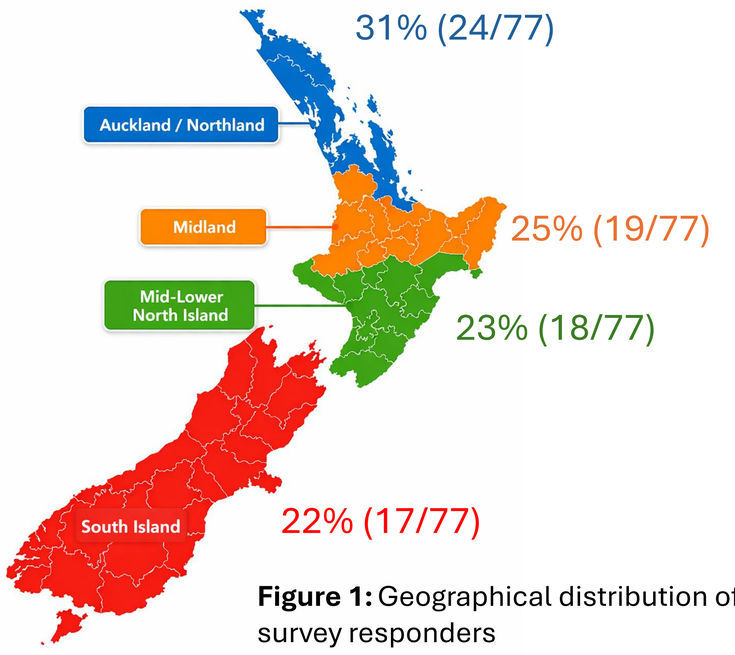

Figure 1: Geographical distribution of survey responders

Response rate and geographical distribution

- 32% (77/240) total responses
- According to speciality
  - 25% (33/130) ORL
  - 27% (11/41) VAS
  - 48% (33/69) ENDO

Results

Estimated numbers of patients with HNPGL seen by survey participants

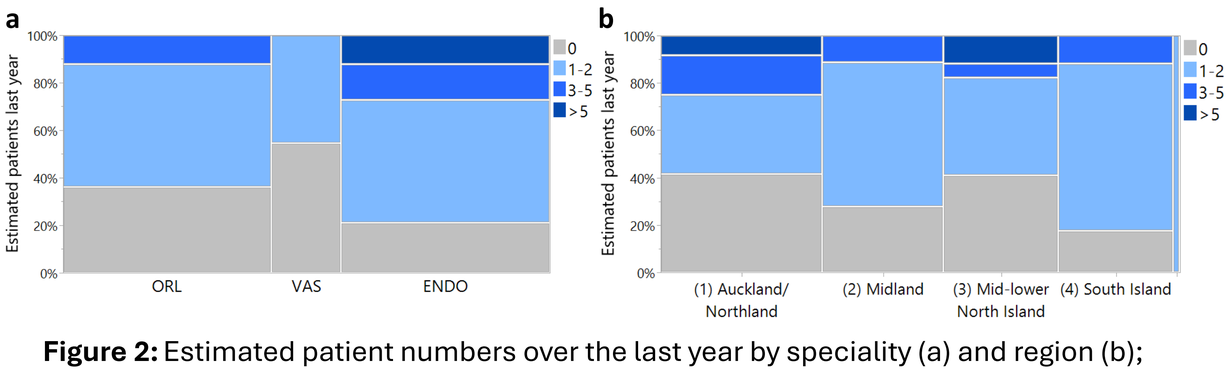

Figure 2: Estimated patient numbers over the last year by speciality (a) and region (b); n=77.

Patients with *SDHx* mutations seen by all specialities across the country

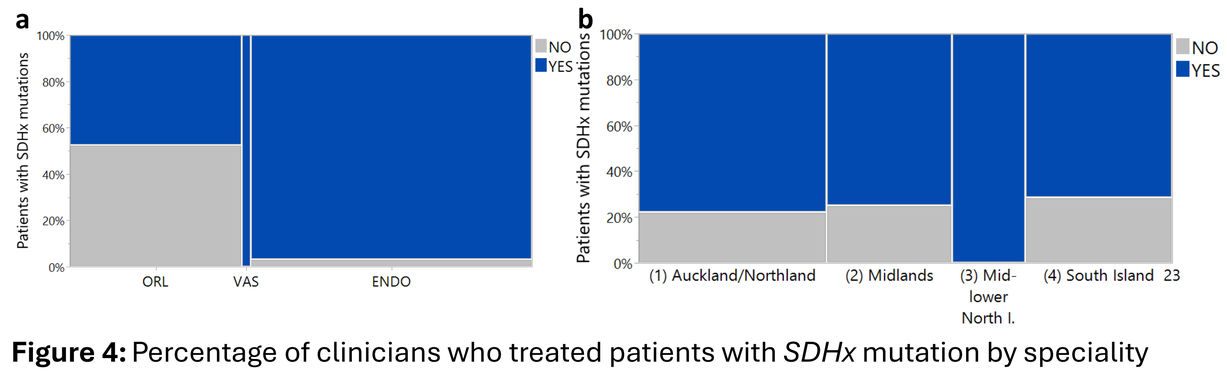

Figure 4: Percentage of clinicians who treated patients with *SDHx* mutation by speciality (a) and region (b); n=51.

Referral rates for genetic testing

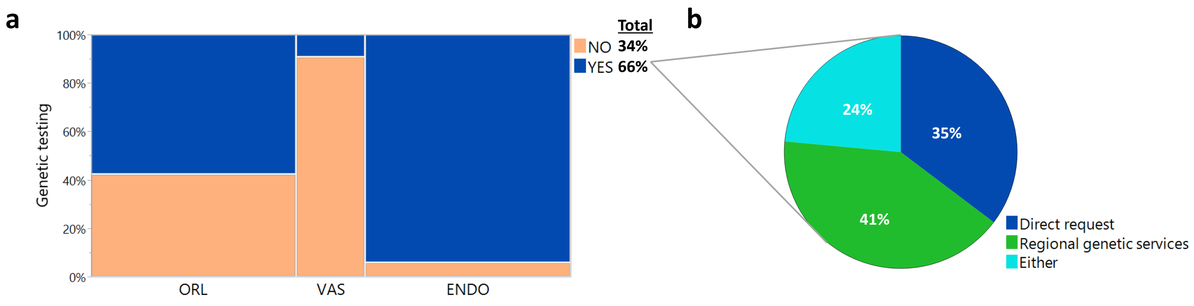

Figure 3: Referral rates to genetic testing by speciality (a; n=77) and arrangement modalities (b; n=51).

Referral to endocrinology for surveillance of mutation carriers

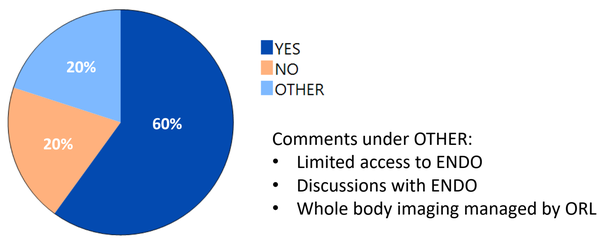

Figure 5: Referral of patients with *SDHx* mutation to ENDO by ORL/VAS speciality; n=10.

Discussion

- **Inconsistent care** across NZ for patients with HNPGL **without clear referral path for genetic testing** and for subsequent surveillance by endocrinologists for patients with *SDHx* germline mutations
- Patients with ***SDHx* gene mutations may be underdiagnosed**, missing an opportunity for early diagnosis and treatment
- Do we need New Zealand guidelines?