“Rebooting” cancer genetic counselling appointments – an 8-week review

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Introduction

> During the period of COVID-19 restrictions in Canberra, the number of patients seen within the ACT Genetic Service dropped. Further compounding already long waiting times (Bates et al. 2021).

> The ACT Genetic Service acquired funding under Operation Reboot, a $5.5 million initiative developed by the ACT Government to reboot outpatients and provide capacity to address the large backlog of long wait referrals (Canberra Health Services, 2020).

> The funding was used to employ a part-time Associate Genetic Counsellor for 8 weeks with the aim of consenting 47 patients for cancer genetic testing via telephone.

> A review of the patients seen within 8 weeks of Operation Reboot was undertaken to inform future practice within the ACT Genetic Service and to provide other health professionals insight into our scope of practice within cancer genetics.

Explainer box

> Predictive testing can be offered if a first-degree family member (or second-degree in certain situations) has had a pathogenic genetic variant identified. This family member would have a 50% risk of having the same pathogenic variant (less if a second-degree family member) and would therefore be at risk of developing the associated cancer. Predictive testing is covered by Medicare.

> Diagnostic testing is gene panel testing which can be considered in individuals who have already been affected by cancer. An example of a gene panel is the BRCA Plus Panel for breast cancer predisposition, which looks for genetic changes in the genes BRCA1, BRCA2, TP53, PALB2, CHEK2, ATM, RAD51C and RAD51D. Diagnostic testing can be covered by Medicare if eligibility criteria is met (chance of finding a pathogenic variant is >10%).

> The approach to cancer patient flow at the ACT Genetic Service is visualised in Figure 1.

Methods

> Patients seen during Operation Reboot (December 2020 – January 2021) were included.

> Data extracted included specialty of referring doctor, category of cancer referred for, type of testing offered, results and whether follow up was required.

> Descriptive statistics were used to analyse the data.

Results

> 50 patients were seen during the study period.

> The three most common referring specialties were General Practitioners (n=25), 50% Medical Oncologists (n=12, 24%) and Endocrinologists (n=6, 12%). Other referring specialists included Surgeons (n=4, 8%), Radiation Oncologists (n=3, 6%), Gastroenterologists (n=1, 2%) and Paediatricians (n=1, 2%) (Figure 2).

> The majority of cancer genetic testing were for breast and ovarian cancers (n=29, 58%), followed by colorectal cancers (n=8, 16%), endocrine cancers (n=8, 16%), prostate cancers (n=3, 6%), endometrial cancers (n=1, 2%) and renal cancers (n=1, 2%) (Figure 3).

> Half (n=25) of the referrals were for consideration of diagnostic genetic testing and the remaining 50% were for the discussion of the option of predictive testing (n=25).

> 80% (n=40) of patients proceeded with genetic testing. 18% (n=9) did not return genetic testing consent forms and 2% (n=1) decided not to proceed with testing.

> Out of the 40 patients who had consented to genetic testing, 32 results had been returned at the time of the audit (80%).

> Of the patients who had predictive testing, 29% (n=5) had a positive result, and 71% had a negative result (n=12).

> Of the patients who had diagnostic testing, 95% (n=14) of patients had no pathogenic variant identified and 7% of patients (n=1) had a pathogenic variant identified (BRCA2 gene).

> Of the patients who had genetic testing results returned (n=32), 28% (n=9) required follow up with our offsite cancer geneticist post result feedback, either for positive results (n=1, 6%) or for management of high risk despite no pathogenic variants found (n=3, 33%) (Figure 4).

Conclusions

> Genetic counsellors within the ACT Genetic Service provide personalised cancer risk information for patients which in turn inform valuable preventative management recommendations.

> Undertaking the genetic counselling appointments over telephone may have contributed to the higher proportion of consent forms not being returned in a timely manner compared to face-to-face appointments (36%).

> The minority (28%) of patients seen by Genetic Counsellors within ACT Genetics were referred for the discussion of cancer risk information for patients which in turn inform valuable preventative management recommendations. Genetic counsellors within the ACT Genetic Service provide personalised cancer risk information for patients which in turn inform valuable preventative management recommendations.