Australian government
Medical Advisory Committee
MSAC.Guidelines@health.gov.au

12 October 2020

Re: Consultation on the Medical Services Advisory Committee (MSAC) Revised Draft Guidelines

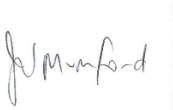
Thank you for the opportunity to provide feedback with regards to the Medical Services Advisory Committee (MSAC) Revised Draft Guidelines

About GUARD
The GUARD Collaborative is a coalition of peak body organisations; Genetic Support Network of Victoria, Genetic Alliance Australia (NSW), Syndromes Without A Name (SWAN) Australia and Genetic and Rare Disease Network (WA). We stand together to represent the voice of people living with genetic, undiagnosed and rare disease and those who support them. We stive for a fair, equitable and collaborative approach to health and wellbeing for all members of our population.

Our submission is in the context of the National Strategic Action Plan for Rare Disease, a focussed plan outlining the priorities and areas of action required to improve the lives of people living with rare disease. We have provided feedback from a patient perspective

I would be happy to provide further information about our submission if required.

Kind regards



Jan Mumford
Director
Genetic Alliance Australia
On behalf of the GUARD Collaborative

RECOMMENDATIONS

The GUARD collaborative would like to make comment and observations for consideration in the development of these Guidelines in support of changes, provision of suggestions and how the changes will impact the genetic rare disease community. The observations are aligned with the National Strategic Action Plan for Rare Disease[1], a focused plan outlining the priorities and areas of action required to improve the lives of people living with rare disease. **Pillar 2 Care and Support** describes the aspects relating to access of treatments.

PILLAR 2: CARE AND SUPPORT

- **Priority 2.1:** Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.
- **Priority 2.2:** Ensure diagnosis of a rare disease is timely and accurate.
- **Priority 2.3:** Facilitate increased reproductive confidence.
- **Priority 2.4:** Enable all Australians to have equitable access to the best available health technology.
- **Priority 2.5:** Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

The provision of personal utility aligns with Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.

The use of personal utility as a new inclusion for the Guidelines is esteemed by the rare disease community. The value of 'knowing' provides a clinical and psychological reference point facilitating treatment, reducing the diagnostic odyssey and improving mental health. The risks are also acknowledged; survivor guilt and challenges to family narratives of health and parentage for inherited conditions.

Personal Utility fits with the biopsychosocial model of health. Work by Borrell-Carrio (2004) proposes clinical practice with

> *a biopsychosocial-oriented clinical practice whose pillars include (1) self-awareness; (2) active cultivation of trust; (3) an emotional style characterized by empathic curiosity; (4) self-calibration as a way to reduce bias; (5) educating the emotions to assist with diagnosis and forming therapeutic relationships; (6) using informed intuition; and (7) communicating clinical evidence to foster dialogue, not just the mechanical application of protocol*.

This approach facilitates a patient- clinical relationship in line with western contemporary medical practice. Personal utility may take other dimensions for Cultural and Linguistic Diverse (CALD) communities, necessitating increased cultural competencies by clinicians and flexibility of service delivery.

Metrics by the use of DALY measures are used for health economics and clinicial utility has detractors and criticisms of this methodology, as it limited to economic productivity dimension. GUARD acknowledges metrics on personal utility are difficult to determine, however indirect impacts to the community metrics can be found. An example of this is community response to fund research for cancer, heart and childhood conditions. Data can be obtained on early access to superannuation as a result of life limiting conditions. The value of personal utility translates to community utility and social capital.

[1] https://rva.blob.core.windows.net/assets/uploads/files/NationalStrategicAPRD.pdf

The Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate and **Priority 2.3: Facilitate increased reproductive confidence** is paramount in harms and benefits deliberations as noted in TG28. Accurate information is inestimable, combined with a limited time frame for options on life affecting decisions, for parent(s) with a foetus with a genetic condition.

Priority 2.2 also applies to those with chronic, debilitating or life limiting conditions and decision/ choices on employment, residence, education, relationships and broader life planning as noted in TG 28. TG 12 notes that impacts to workforce should not be considered in the assessment as a deterrent to a positive assessment. However, the increased service needs by genetic counsellors have been noted in government reports but limited increase in workforce is evident by the delays in patients obtaining a consultation. This increases mental stress for the patient and genetic counsellors. Failure to deliver timely and accurate information will reduce confidence in genomic technology and Australian health services resulting in poorer health outcomes and ripple effects for all Australians.

Priority 2.4: Enable all Australians to have equitable access to the best available health technology. The use of Patient Reported Outcome Measure's (PROMS) is noted and valued by consumers. Data can be collected in all trial stages and this information shows significant benefit, both with high and low clinical utility. However, key characteristics of rare diseases is that the knowledge base available is very often limited and limited knowledge of responses to health technologies places additional onus on PROMS. Clinical trials design for rare conditions have a small population and a smaller suitable trial cohort to draw from necessitating innovative trial design, such as basket trials, and pioneering statistical methods for validity. International bodies such as the International Rare Diseases Research Consortium (IRDiRC) have recommendations for trial design for this community.

Data collection can be by various media- paper questionnaires, electronic questionnaires and personal wearable devices. A strength of electronic data collection is the facilitation of rapid collection of data, reduces recall bias and facilitates quicker recognition of adverse events, thereby increasing patient safety. The use of electronic data collection will enhance participation in trials in regional and remote, provision for languages other than English will facilitate participation from our diverse communities within Australia. The use of electronic PROMS notably the arena of wearable device technology is growing and is not noted in the Guidelines. This gap needs to be addressed as it reduces data collection and effective patient management. By recognising incoming technology, new and novel health technologies, if safe and suitable, can be introduced quicker for community consumption.

Additionally, repurposing of health technologies from mainstream or common conditions may be done for rare conditions under 'rule of rescue' combined with PROMs to guide safety and efficacy measures.

Work in understanding the psychological issues and significance aligns with the Strategic Plan **Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support.**

The Guidelines cite examples from cancer literature and inherited conditions, but not *de novo* conditions which affect many families. The psychological impacts of unclear results for *de novo* conditions is very real for families where testing does not provide a diagnosis to account for a child's

condition. This is increased as data on psychological impacts of genomic testing is scattered. A gap exists as the psychological impacts of genomic technology, as no professional body exists in Australia to provide support and research data on the unique stressors of genetic testing, social impacts or guidance to provide education or reduce stress.

Psychological aspect of cancer diagnosis and treatments is well researched by the area of psycho-oncology. Genetic Counsellors provide service in assisting patients but for acutely distressed patients, understanding of family narratives and those facing significant life decision such as termination or to continue a pregnancy. This gap in knowledge contributes to ethical and moral debates on suitability of services offered and funded by the Australian Government. Work on a knowledge base reflecting the Australian context is needed to best inform decisions on mental health, social norms and more and the emotional wellbeing of those affected directly and indirectly by rare conditions.

Genetic testing should have an accompanying care pathway communicated to the person, or authorised guardian, requiring the testing. Ideally a dedicated Nurse Care Coordinator should be assigned to each person to assist with navigating the health system for complex conditions.

Broadly, the following also applies.

The epidemiological approach may not be suitable for rare disease. The Review notes on page 159, market growth can be used to predict increase in patient numbers. An increase is expected for rare genetic conditions, but with an unknown base as there are no mandated reporting or database of rare conditions in Australia. The expected growth and use of genomic technology will increase diagnosis of rare conditions, but numerically difficult to predict. Extrapolation from overseas may be used but may not apply to the Australian context due to migration patterns or the Australian First Peoples.

Further, internationally the number of therapies for rare diseases has increased dramatically in the past decade and will continue to increase in the foreseeable future. People living with a rare disease must be able to hope for access to new drugs and treatments in Australia and to benefit from them rapidly and fully, all across world, as soon as they are approved.

TG 6.3 titled ‘Other Approaches’ may benefit from an alternative title, such as ‘Less common conditions and treatments’. The strategies suggested here would best apply to less researched areas of health such as rare conditions. With the growth of genomic technology and the demand for effective treatments, clear signposting within the guidelines would signify commitment to health equity for the rare community.

GUARD would like to acknowledge that support for patients and patient support groups, who have limited time, capacity and energy, improved easier communication and collaborative work solutions are needed to introduce new treatments.