rare voices
AUSTRALIA

24th August 2015

Dear PBAC Guidelines Review Secretariat,

Re: PBAC Guidelines Review Public Consultation

Rare Voices Australia Ltd. (RVA) welcomes the opportunity to respond and provide input and its views to the PBAC Guidelines Review Public Consultation. It is understood the review will be considering a range of items in detail (as outlined in detail on the Department of Health website; 'Items to be included in the Guidelines Review' items 1-6). Abbreviating the items as follows;

1. Review of the Guidelines content identifying the methods and current research. Identify significant new developments for methods in relevant sections of parts II and III of the current Guidelines.
2. Preparation of a technical paper for discussion on each issue for the revision, including identifying any issues of scientific debate and consideration of Australian and International best practice.
3. Consolidation of these into a revised set of Guidelines for methods (with any matters related to process identified and agreed as out-of-scope).
4. Management of a public consultation process for the proposed revised Guidelines for methods.
5. Incorporate accepted consultation feedback into the draft revised Guidelines.
6. Present the finalised revised Guidelines for consideration by the DUSC and the Economics Sub Committee ESC, and for endorsement by the PBAC.

Rare Voices Australia Position

RVA is keen to see the PBAC Guidelines strengthened by creating efficiencies in process, representation for the rare disease sector from a range of stakeholders including the rare disease health consumer and consider the rare disease principles and objectives to enable safe and effective treatments to be readily accessible to the rare disease community. People living with a rare disease live in the hope of one day a treatment will be discovered and that the process is a fair, equitable and transparent one with efficiencies in place to ensure treatment listing is efficient and without delay.

Rare Voices Australia Background

Rare Voices Australia, the National Alliance of the rare disease community is a unified voice of an estimated 1.2 - 2 million Australians living with a rare disease. RVA members compromises of over 200 rare disease patient organisations and more than 2000 members throughout Australia including adults and children living with a rare disease.

RVA's Mission; the national alliance of people living with a rare disease, will provide a unified voice to improve the lives of all Australians affected by rare diseases.

RVA Vision: Australians with rare diseases will have extended and improved lives. Access to world class, improved, better, best practice in areas of; diagnosis, treatment, care, services, support services and therapies. Reducing mortality and morbidity enabling the 'best' life possible.

1. Review of the Guidelines content identifying the methods and current research.

Guidelines for preparing submissions to the Pharmaceutical Benefits Advisory Committee is essential and necessary to ensure treatments listed are safe and effective for Australian health consumers. The PBAC Guidelines could be strengthened and incorporate specific guidelines that meet needs of rare diseases. Unlike common conditions, strong robust data sets is difficult to find especially as Australia does not collect data for rare diseases officially. RVA recommends that the PBAC Guidelines consider the nature of rare disease and how to manage risk and manage small data sets from across local and international sources. The methods currently used could consider incorporating knowledge, methods and current research utilised for rare disease in UK and Europe.

2. Preparation of a technical paper for discussion on each issue for the revision, including identifying any issues of scientific debate and consideration of Australian and International best practice.
Without citing the technical paper it is difficult to make comment. RVA recommends that the paper considers data from both local and international sources. In Australia whereby local knowledge in rare diseases is often scarce and scattered, the level of expertise will be limited. RVA recommends that the PBAC look at how this is managed in UK and in EU in particular for rare disease and processes used to include and identify any issues of scientific debate.

3. Consolidation of these into a revised set of Guidelines for methods (with any matters related to process identified and agreed as out-of-scope).
The PBAC process has recently has taken into consideration rare diseases and explored new 'out of scope' processes to further understand new innovative treatments and how best to review at a PBAC level. RVA proposes that the PBAC adopt a formal process for listing rare disease treatments that considers the need to allow efficiency in process avoiding lengthy time delays. A process whereby PBAC directs a stakeholder meeting to consider the listing of a new rare disease treatment with equal representation from a range of stakeholders; rare disease clinical / research experts, PBAC Director/ representative, TGA representative, Sponsor company representative, Rare Disease patient organisation / RVA representative. To date this has been trialled by former PBAC Director and recently by Department of Health under the sponsor company direction. The process by which this occurs should be consistent across all rare diseases and the facilitation should be from PBAC with agenda, minutes and outcomes made transparent to all involved.

The stakeholder meeting allows all relevant materials to be reviewed, clarification of data and questions raised and answered in a collaborative environment. Key outcome to bring all necessary data to PBAC to consider and make its decision based on information from local and international data. As part of best practice, the meetings need to state objectives, outcomes and minute discussions. All to be made available for public review.

This process will enable and address the time delays for resubmitting to PBAC if a decision is deferred or rejected. Whereby additional data is required, the process and manner in which this is addressed again needs to take into consideration the burden that process has not only on the Sponsor Company but the patients themselves. The key outcome is to improve inefficiencies in a system that currently does not formally consider rare disease treatments. Thus looking at how to improve process which ultimately improves health outcomes for health consumers in Australia.

4. Management of a public consultation process for the proposed revised Guidelines for methods.
RVA supports a public consultation process for the proposed revised Guidelines for methods and would like to see that other international processes for listing rare disease treatments used in UK and Europe be revised and considered as part of the scope of work.

5. Incorporate accepted consultation feedback into the draft revised Guidelines.
RVA recommends that rare diseases are considered in a different light to that of common conditions. Some important points to consider that are shared amongst the many variety of rare diseases include;

I. **Lack of access to correct diagnosis**
The specific and challenging characteristics of RD result in patients experiencing late or inaccurate diagnosis, fragmented access to care and inadequate or inappropriate treatment proving both harmful and of great risk to the RD patient.

II. **Lack of information**
Dissemination of scarce existing information is a key issue for the rare disease community at large.

III. Lack of scientific knowledge
This results in difficulties in the areas of therapeutic tools development, definition of the therapeutic strategy and in shortage of therapeutic products, both medicinal products and appropriate medical devices

IV. Social consequences
Living with a RD has implications on all areas of life; school, work choice, leisure time are all affected leading to isolation, exclusion from social community, discrimination for insurance (health, travel and mortgage insurance) and reduced professional opportunities.

V. Lack of appropriate quality healthcare
Appropriate healthcare would entail combining the different spheres of expertise needed for rare disease patients and adopting appropriate models of care. The “Multidisciplinary” clinic models have been adopted for some of the rare diseases in Australia and have resulted in treatments being listed on either the Life Saving Drugs Program or PBS (Pharmaceutical Benefits Scheme).

VI. High cost of the few existing drugs and care
The additional expense of coping with the disease (human, medical and technical aids), combined with the lack of social benefits and reimbursement, always leads to an overall pauperisation of the family, and dramatically increases the inequity of access to care for rare disease patients. Most often families simply cannot afford the best available healthcare for their loved ones.

VII. Inequities in availability of treatment and care
Innovative treatments are unevenly available across the world including Australia. Clear contributing factors include; economy, scarce priority given to rare diseases within the Public Health sphere. Additionally delays such as; price determination and/or reimbursement decision, lack of experience of the treating physicians (not enough physicians involved in rare diseases clinical trials), and the absence of treatment consensus recommendations in Australia and globally is an ongoing problem.

6. Present the finalised revised Guidelines for consideration by the DUSC and the Economics Sub Committee ESC, and for endorsement by the PBAC.

RVA would like to see a discussion paper with draft revised Guidelines made available to review and make comment. PBAC Guidelines needs to consider the various sub-sets and groups in need of access to medicines and treatments for their condition.

Rare Disease Treatments need to be Accessible via a seamless and efficient process

Rare Disease treatments need to be accessible. The process by which they are listed and reviewed needs to recognise the people it is set up to serve being the people who are impacted each day living with a chronic, life threatening, debilitating condition whereby no other treatment option is available to them and the treatment will potentially bring improvement to their quality of life.

According to the report; ‘Funding Rare Disease Therapies in Australia Ensuring Equitable Access to healthcare for all Australians’ (The McKell Institute), there are inefficiencies experienced in Australia compared to that of international comparable countries with regard to accessing therapies for rare diseases. (1) The average orphan drug marketing application is taking on average 15 months to be approved including the pre-submission process. In comparison to the United States and Europe the orphan drug marketing applications are evaluated in 10 months or less.

There is opportunity for the PBAC to further streamline process and speed access to innovative orphan drugs for rare diseases.

Principles

RVA recommends that some shared Principles that underline rare disease be adopted and linked to the Orphan Drug Program for Rare disease and considered as part of the review of the PBAC Guidelines.

1. Social Justice, Equity and Solidarity principles

Based on the principles of social justice, equity and solidarity, the needs of rare disease populations have to be specifically addressed, as they should be for any minority or underserved community. This is an important part of the goal of reducing health inequalities for rare diseases in Australia.

People living with a RD are a major healthcare concern and unlike other key health priorities they are not the result of 'poor lifestyle choice i.e. smoking, drinking.' Australians living with a RD are born with their RD and the disability that also is associated with it.

The UN Convention on the Rights of the Child (1989) includes right to the highest attainable standard of health care.

The Australian Human Rights Act (2) and disability discrimination legislation (3) promotes the right to enjoy a life as normal and full as possible, the right to medical treatment, the right to measures that will enable individuals to become as self-reliant as possible, and the right to have their special needs taken into consideration at all stages of economic planning.

While it is acceptable that the implementation of these instruments is subject to the laws, policies and budgets of its jurisdiction, it is not acceptable that public health planning and services often give so little priority to the needs of rare diseases patients. The consequence of this lack of interest has been to reinforce the disadvantages of people living with rare conditions in Australia.

Accessing a medicine that is safe and effective for someone with a smoking addiction (a lifestyle choice common health concern) is not the same as listing a treatment for someone who has been born with their very rare, life threatening, debilitating and life limiting disease. It is very difficult to make direct comparisons and therefore the processes for listing treatments and ensuring they are accessible needs to reflect this principle.

The EU Regulation on Orphan Medicinal Products (4) is an example of rare disease research translating into effective outcomes and a marked 'true success' by European standards. In Europe over the last 10 years, only approximately 100 new RD medicines – with an orphan drug designation or not – have been approved in the EU2. The number of RD projects funded by the European Commission Directorate General for Research Framework Programs (FP), in particular FP6 and FP7, is steadily growing.

Approximately 5,000 research projects in EU currently cover 2,000 different RDs. In addition over 650 clinical trials are ongoing for more than 300 RD (including for designated orphan drugs). However, there are thousands of RD for which there is no ongoing research activity.

In particular, for RD with a prevalence of <1/10,000, therapeutic research is very limited or non-existent. The Commission Communication on Rare Diseases proposes that Member States put in place strategies to foster RD research, including cross-border cooperation and collaboration to maximize scientific resources across the EU. (5)

The Commission's Council Recommendation recommends that Member States establish and implement plans or strategies to ensure provisions aimed at fostering research in the field of RD. Both documents call for a RDR policy which is both comprehensive (covering a large scope from basic to clinical research) and integrated (EU and national levels). The European Project for Rare Diseases National Plans Development (EUROPLAN) further specifies guidelines and recommendations for the elaboration of national action for RDR in the context of a national plan. (6) The E-RARE project provides an important forum for Member states to exchange about their respective RDR policy and to coordinate their strategies.

Conclusion

Rare Voices Australia would like to acknowledge the TGA, PBAC, Department of Health and other department's willingness to engage with RVA allowing the patient community voice to be included and heard in the *PBAC Guidelines Review Public Consultation*.

Australian patients living with rare diseases call for the Australian Government to adopt a National Rare Disease Strategy and it is pleasing to report that the Western Australian Department of Health have now published a Western Australian Rare Diseases Framework 2015-2018 endorsed by WA Director General of Health. Rare Disease patients need to access treatments that have been approved by the TGA as safe and effective in a system that adheres to a transparent, efficient and equitable process. They need to have confidence in the PBAC who understand rare diseases and importance of access to safe effective therapies and treatments. Most of these medicines and therapies are innovative, new and some are complimentary or in addition to existing medicines or products. Some rare disease treatments are specifically targeted towards very rare diseases. There needs to be flexibility in the approach to listing such treatments and guidelines drafted need to consider this carefully to ensure patients who can benefit from accessing them, will in fact receive them in a timely manner to optimise opportunities to show and receive clinical long term benefit.

Kindest Regards,

MFookes

Megan Fookes
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Rare Voices Australia Ltd.

References

1. McKell Institute Report, Funding Rare Disease Therapies in Australia, Nov 2014, http://mckellinstitute.org.au/wp-content/uploads/pdf/McKell_RareDiseases_A4-New-web.pdf
2. *Australian Human Rights Commission Act*. 1986: Australian Government
3. *Disability Discrimination Act*. 1992: Australian Government
4. EC Regulation 141/2000 on Orphan Medicinal Products
5. COM (2008) 679 Communication from the Commission to the European Parliament, the Council the Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges.
6. *Final recommendations available on the EUROPLAN website, www.europlanproject.eu*

Appendix

RARE DISEASES SUMMIT - DRAFT COMMUNIQUE

Principles and objectives to progress a National Plan for Rare Diseases

1. **Rare diseases need to be recognised as a national health priority.** The burden of rare diseases, while largely hidden due to inadequate information systems, is unacceptably high for patients, families and the community.[1] People living with a rare disease have shared concerns and needs that warrant national recognition, leadership and action.
2. **More equitable and timely access to diagnostics, treatments, services and coordinated care for people living with a rare disease.** It is in the national and state interest to establish leadership, direction and action towards more effective national systems and approaches to rare diseases. Appropriate service models should be identified and harmonised across states and territories and developed for regional and **Incentives are required to drive a coordinated and collaborative action on rare diseases. A national approach should be established, backed by the Australian government.** There is no shortage of opportunities to improve diagnostics, treatments, services and coordinated care. A challenge is to reach agreement on how best to focus effort. Incentives should bridge efforts at local, state, national and international scales and support integrative approaches which leverage knowledge, skills and capacity.
3. **Leaders from patient, healthcare, research, and government and industry organisations need to work collaboratively to champion a National Plan.** The Plan needs endorsement from all of the Australian, state and territory governments. A National Plan requires agreement among the key stakeholders.
4. **The National Plan should identify a set of objectives and enabling strategies to drive action.** Six objectives are proposed under key themes;
    - i. **National Coordinated and Collaborative Approach**
      Establish a national platform for dialogue, information exchange, and coordinated and collaborative action on rare diseases.
    - ii. **Data Collection and Use**
      Identify and enable approaches to data collection and use that better meet the needs of people living with a rare disease.
    - iii. **Coordinated Care**
      Identify and enable approaches to coordinated care that better meet the needs of people living with a rare disease.
    - iv. **Equitable Access to Services**
      Ensure Australia's health system enables timely, equitable access to appropriate services for people living with a rare disease.
    - v. **Equitable Access to Diagnostics and Treatments**
      Ensure Australia's health system enables the development of diagnostics and treatments for rare diseases and facilitates timely, equitable access for patients.
    - vi. **Nationally Coordinated Research**
      Establish a nationally coordinated program of research on rare diseases that is informed by patients and carers.

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[1] An estimated 1.2 -2 million Australians live with a rare disease.

Endorsements are still being received at the date of this publication – 24.8.15

Organisational endorsement of the Rare Disease Summit Communique

amdf
AUSTRALIAN MITOCHONDRIAL DISEASE FOUNDATION

apsu
Australian Paediatric Surveillance Unit

ausEE

pompe
Australian Pompe's Association

AWCH
association for the wellbeing of children in healthcare

BIOMARIN

CYSTIC FIBROSIS ACT

Duchenne Foundation AUSTRALIA
Strength Through Support

ECD
ERDHEIM-CHESTER DISEASE GLOBAL ALLIANCE

Fabry support group Australia

GAA
Gaucher Association of Australia

Gaucher Association Australia

GENETIC AND RARE DISEASE NETWORK

genzyme
A SANOFI COMPANY

HAE Australasia
hope • advocacy • education
Australasian Patient Organisation for Hereditary Angioedema

HCU Network Australia
HCUNetworkAustralia.org.au

Endorsements are still being received at the date of this publication – 27/4/15

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Self Help
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FAST
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CUREAngelman.org

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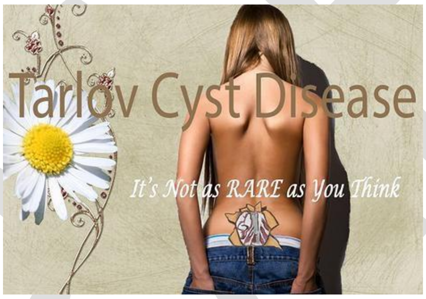

GELASTIC SEIZURE SUPPORT HUB
"IT'S NO LAUGHING MATTER!"

QLD
Carers
AUSTRALIA
Placing Carers First

Australian CYSTINOSIS Support Group

PALLISTER-HALL SYNDROME (PHS) SUPPORT HUB
KNOWLEDGE = CHOICES = STRENGTH

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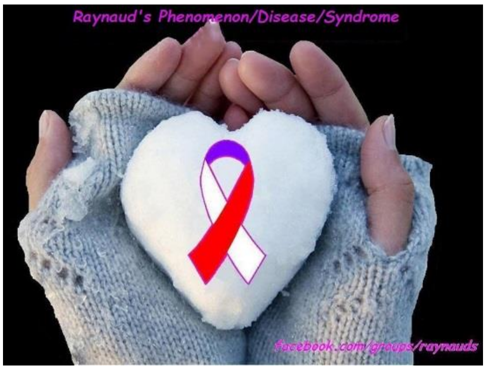
New South Wales

CYSTIC FIBROSIS
Tasmania

Tuberous Sclerosis
Australia



MdDS Australia

Finding a Balance Through

- Support
- Friendship
- Awareness
- Research

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