Pfizer Australia welcomes the opportunity to provide a submission to the review of the Life Saving Drugs Programme (LSDP) and applauds the Government for conducting this review. While the system of funding new therapies to treat rare diseases is in need of reform, it is only through the LSDP, or an equivalent funding source for medicines for rare diseases, that treatments can be made available for patients with rare diseases.

## Introduction

Pfizer has considerable experience in, and a strong commitment to, the treatment of rare diseases. The term rare diseases is used to cover a wide variety of conditions, including neurological disorders, infectious diseases, rare cancers, autoimmune disorders, respiratory diseases and a variety of genetic disorders. Some rare diseases affect only a handful of patients around the world, while others may affect 200,000 or more. Over half of all rare diseases affect children, and more than 3,000 have a known genetic component.

Pfizer has more than 22 medicines approved worldwide for the treatment of rare diseases, including haemophilia A and haemophilia B, acromegaly, pulmonary arterial hypertension (PAH), transthyretin-related familial amyloid polyneuropathy (TTR-FAP), Gaucher disease, and rare cancers, such as gastrointestinal stromal tumour (GIST) and ALK-positive non-small cell lung cancer. In addition, our current pipeline includes clinical and pre-clinical programs in sickle cell disease, haemophilia, muscular dystrophies, spinal muscular atrophy, cystic fibrosis, and a number of rare cancers and patient subpopulations classified as rare diseases.

Through the innovative work of researchers, including those supported directly and indirectly by pharmaceutical companies such as Pfizer, opportunities to develop therapies to treat previously untreatable rare diseases are emerging. It is critical that the health funding environment is appropriately structured to allow Australian clinicians and patients to benefit from these developments.

Innovative science is leading to an improved understanding of the genetic basis of many diseases, coupled with advances in biotechnology and drug development. This is leading to improved treatment options for patients, and increased expectations about the capacity of medical advance to improve health outcomes. However, the reality is that the rarity of these conditions means that clinicians have very limited experience with them, and that many rare disease patients wait years before receiving an accurate diagnosis. Newborn screening programs and approved, validated genetic tests can all help speed the process for many genetically-linked rare diseases but many rare disease patients undergo a "diagnostic odyssey," involving many different clinicians and hospitals and lasting many years before their condition is properly identified. Even then, treatment options may be limited or non-existent: fewer than five percent of all rare diseases have approved therapies available. Due to the paucity of approved therapies, medicines are sometimes used off-label to treat rare diseases.

### A challenging area of health

New orphan drugs have allowed patients with rare diseases such as Gaucher disease and haemophilia to live longer and have a higher quality of life. There can be little doubt that continued progress in other rare diseases where no current approved therapies exist would

transform lives and give physicians much needed treatment options for their patients. The reality is that, left untreated, rare diseases can generate significant morbidity and mortality and impose major burdens on health and social care systems. Enabling patients to have timely access to appropriate medicines is therefore a social as well as economic imperative.

However, developing therapies for rare diseases poses many unique challenges. There is usually little understanding or information about the natural progression of the disease to inform trial design, and investigators often have difficulty identifying and enrolling a large number of patients. Basic research tools, such as validated animal models, may not exist. These challenges increase the uncertainty that a research program will lead to a new therapy. Coupled with small patient numbers and, hence, limited potential revenues, there has historically been less investment into rare diseases than into more common diseases.

In recognition of these specific issues facing drug development for rare diseases, many governments around the world have developed orphan drug regulations to support the development of new products intended for the diagnosis, prevention or treatment of rare conditions. While provisions vary from country to country, the key incentives created under various orphan drug regulations generally include marketing exclusivity and support for sponsors taking their orphan drug through the regulatory approval process. Some regulations also include research grants or R&D tax credits.

## The review

The LSDP review is a timely opportunity to ensure Australia develops a funding arrangement that will support continued innovative research by ensuring patients are able to benefit from ongoing medicines development.

This review is necessary to ensure that Australians with rare diseases are not disadvantaged by the inability to access medications necessary to stabilise their conditions, extend their life and improve their overall quality of life. Equity dictates that they should have equal ability to obtain medications to persons who have their treatments for more common conditions readily available through the Pharmaceutical Benefits Scheme.

It is acknowledged that there are evidentiary limitations due to the small populations involved in rare and ultra-rare conditions. Additionally, submissions for these products are unable to meet the rigorous cost-effectiveness criteria of pharmaceuticals to be listed on the Pharmaceutical Benefits Scheme (PBS) with incremental cost-effectiveness ratios for these products being significantly higher than would be acceptable to the Pharmaceutical Benefits Advisory Committee (PBAC). Nevertheless these medicines should be reimbursed in a timely manner to meet the tenets of the National Medicines Policy which includes: "Timely access to the medicines that Australians need, at a cost individuals and the community can afford."

The LSDP review webpage indicates that: ".... it is an opportunity to review the current programme in order to ensure that Australians with very rare conditions continue to have subsidised access to much-needed, expensive medicines."[1] In fact, the inclusion of "continue"

[1] http://www.pbs.gov.au/info/reviews/life-saving-drugs

in this statement is inaccurate. Currently, there are nine medicines funded for seven rare diseases under the LSDP. Since May 2010, there have been 17 submissions (of which 10 were resubmissions) for six medicines for seven conditions, six of which currently do not have any reimbursed treatments. Only two of the medicines which were the subject of these submissions are now reimbursed under the LSDP. Therefore, although continued treatment of patients currently receiving medicines under the LSDP is crucial, a focus should also be on ensuring access of treatment for patients for whom treatments are currently not being funded.

As a consequence of the current process, Australians are being denied access to new medicines which are funded in other countries and when medicines are funded, patients have to wait far longer to access treatment.

In considering an appropriate future structure for the funding of medicines for rare diseases, and the future of the LSDP, Pfizer encourages the committee to have a view to the following:

- Health Technology Assessments for orphan drugs needs to capture the full societal benefit of new treatments for rare diseases. They require different methodologies from the assessments designed to evaluate medicines for common diseases. Despite the very small patient populations, the reality is that rare diseases can generate significant morbidity and mortality and impose major burdens on health and social care systems.
- Funding and regulatory evaluations need to be recognised and support the specific issues facing innovative research into treatments for rare diseases.
- In order to improve patient outcomes, we need to prioritise the early and accurate diagnosis of serious rare diseases, and support patient access to diagnostic testing, as well as centres of expertise in treatment.
- In addition, it is important that there be continued funding for medicines to treat rare diseases, in part to maintain investment into the vital research that will lead to new rare disease therapies. In this area, appropriate intellectual property and tax incentives to encourage the development and commercialisation of orphan drugs is critical.

## Need for approach for funding of medicines for rare diseases

There is no doubt that a new approach for the funding of new medicines for rare diseases is required. The current process, particularly since the changes to the process in May 2010, has meant that many patients with rare diseases do not have treatment options.

It is acknowledged that the PBAC is the appropriate body to assess products for funding under the LSDP. However, a specific flexible process should be available to allow timely access to medicines that treat rare diseases.

The current system, which assesses these medicines against PBS-listing criteria, rejects them on cost-effectiveness grounds, and then considers whether they meet the criteria for the LSDP seems illogical. Parallel evaluations should be in place with different sets of criteria for the PBS and the LSDP, with more flexible criteria for the LSDP.

The process for consideration of rare diseases should also have flexibility in terms of clinical evidence in addition to cost-effectiveness. There are a number of issues in conducting clinical studies for patients with rare diseases including: 1) the limited number of patients for enrolment in clinical studies; 2) lack of feasibility of performance of randomised controlled clinically studies, and 3) limitations on inclusion of placebo arms due to ethical issues. This means that the clinical evidence is not of the quality usually expected in submissions to the PBAC.

## Conclusion

In conclusion, Pfizer Australia welcomes the review of the LSDP and looks forward to a reform of the process which will allow access to medicines for patients with rare diseases in a timely manner.