XXXX,

DUBLIN

18th November 2016

Dear Minister Mc Entee,

We are writing to you in your capacity as Minister of State for Mental Health and Older People to request your assistance to highlight the plight of one of your young constituents, our daughter XXX (age 1).

XXX was born in October 2015. Six days after her birth we were rushed into Temple Street after the results of the standard newborn blood screening test were returned. Since then our lives have changed dramatically, as that day, XXX was diagnosed with phenylketonuria (PKU).

We are eternally grateful to the policy makers and clinician from the 1960's who had the foresight to establish this screening programme as without it, XXX would already be severely brain damaged.

PKU is a rare genetic disorder that affects my child's metabolism. People with this disorder are unable to break down an amino acid called phenylalanine (phe). This is a natural substance found in food but in people with PKU, phe builds up in the blood and in the brain and can cause serious problems, including if untreated, severe brain damage and associated psychiatric illnesses.

While PKU is a chronic illness with no cure, there is a way to prevent the build up of phe in the blood and brain. Prevention for people suffering from PKU involves a very challenging, highly restricted, low protein diet.

Living with a *chronic illness* is a constant daily challenge for us as a family and has had a significant impact on our quality of life. Managing XXX's condition offers serious daily challenges relating to her very strict diet restrictions. XXX has a severe strain of classic PKU and is therefore limited to only 4 regular protein exchanges per day (e.g. 1 exchange = 1 level tablespoon of o*a*ts/ 1 teaspoon of lentils/2 level tablespoons of peas) and restricts/prevents the intake of normal everyday protein foods such as: potatoes, breads, pasta, rice, cereals, and so on. Unfortunately, the amount of protein that PKU patients can safely ingest does not increase as they grow. Therefore, we cannot imagine how challenging this will be as she grows as we will have to rely more-or-less <u>completely</u> on the very restricted selection of low-protein foods currently available within Ireland.

As a result of PKU, XXX is also at risk of many other health complications associated with this diet.

Like many other families living with PKU and numerous other rare diseases, I would like, wherever possible, more resources invested in improving our treatment and care. In particular, I want to know that I have access to the full range of low-protein food products (recently seen at PKU International conference in Dublin) that offer my daughter XXX not only the best chance of eating a healthy diet but also a better quality of life.

We fully understand that you are inundated with requests from many constituents related to a myriad of healthcare challenges, and that these inevitably plea for more resources.

XXX has the potential to make an enormous contribution to society throughout her lifespan (optimally treated PKU patients can live long healthy lives). Based on this here are the main issues that we would like to see addressed, or at least engage with you on how best to approach from a policy perspective:

1. The inefficient and antiquated bureaucratic process whereby the range of low- protein products available through the Long Term Illness (LTI) scheme has not kept up-to-date with the obsolescence of some old products, as well as the introduction of new ones. This should easily be rectifiable without undue budget impact.
2. There is medicine called *Kuvan* that has full regulatory approval both in the EU and US since 2007 that can dramatically improve the lives of some PKU patients. However, the HSE refuses to reimburse it, for any patients. As a medical doctor who now works in the pharmaceutical industry I (XXX) am only too aware of the budget constraints that the HSE faces and how this impacts on the introduction of innovative new medicines. And in this particular area I often question the wisdom whereby new medicine reimbursement is often played out in the political arena, rather than the clinical. Our current assessment body, the National Centre for Pharmacoeconomic Evaluation (NCPE) takes the somewhat limited perspective of the *payor*, rather than the *societal* perspective. This inevitably biases approval of new medicines towards those that deliver short-term displacement of existing medical costs, rather than those that deliver greater long-term societal benefits.

Therefore, in due course I also would like to be given the opportunity to discuss with my paediatrician whether the medicine Kuvan (or indeed other future innovative medicines that deliver societal cost-effective benefits) would work for my child, and if so, for it to be reimbursed by the HSE.

3. Finally, to improve significantly the quality of life of PKU people and improve our overall health outcomes, we would like to see a generally more strategic approach to the management of rare diseases in Ireland. We understand that a National Rare Disease Plan[i] (2014-2018) is in place but that there is little transparency or accountability on the progress being made. Any insights you may be able to offer us on how much progress has been made would be greatly appreciated.

We would greatly appreciate the opportunity to meet with you and discuss the above further, so as to enhance our mutual understanding of how, as citizens, we can ensure a better future for XXX, and other PKU sufferers.

We hope that with your help we can optimize outcomes for XXX, inc her psychiatric outcomes, that may be jeopardized without access to appropriate care.

If you require any further detail or information about PKU, please let me know.

Yours Sincerely,

XXX

[i] http://health.gov.ie/wp-content/uploads/2014/07/EditedFile.pdf