Rare Disease Taskforce

General Election Feb 2020 Recommendation of the Rare Disease Taskforce

At the meeting of the Rare Disease Taskforce on Wednesday 15h January 2020, agreement was reached to recommend 3 key asks of every election candidate in the run up to the Irish General Election scheduled for Saturday, 9th February.

#PLEDGE4RD

WE WANT

Budget and resources to implement the Rare Disease Plan

An urgent plan to build adequate Genetic Services in Ireland

Legislation to expand New Born Screening

REMEMBER

- There are an estimated 300,000 people living in Ireland with a rare disease. This means an average of **7,500 individuals and their families living with a rare disease in each of the 39 general election constituencies**.
- A rare disease does not just impact the affected individual. **A rare disease has implications for everyone in the immediate family unit and the wider family too**; grandparents, aunts, uncles, cousins, second cousins.

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Key ask 1 – Budget and resources to implement the Rare Disease Plan

The National Rare Disease Plan for Ireland 2014 – 2018 was adopted as policy by the then Minister of Health, Dr James Reilly, TD in July 2014. This plan remains the policy framework for care of people with rare diseases in Ireland.

- We are calling on all candidates to support full implementation of the policy recommendations through provision of adequate and targeted resources in the next Dáil. This can be facilitated through the inclusion of key elements of the plan in the annual HSE National Service Plan.

Key ask 2 – An urgent plan to build adequate Genetic Services in Ireland

Ireland lags well behind our neighbours in Northern Ireland, the UK and Europe in terms of resourcing of genetic services. The Clinical (Medical) Genetics waiting list was 3,021 on 19/12/2019[1], an increase of 14.5% on the same figure for December 2018[2]. **Over 1/3rd of these patients have been on the waiting list for >1year.** These figures do not include waiting lists for genetic counsellors. It is inhumane to provide a genetic diagnosis over the phone with NO follow up counselling services, as is practice with today's healthcare service.

A failure to provide adequate genetic services means a delay in diagnosis, a lack of appropriate treatment, probable disease progression, and possible misdiagnosis, with associated unnecessary and even harmful intervention. The typical time for diagnosis of a rare disease is 5 years and involves 7 separate specialist consultants, only adding to our waiting lists.

A diagnosis with an inheritable condition has immediate implications the wider family (typically 64 people in the wider 3-generation Irish family) and for family planning.

- We are calling for immediate provision of adequate resources to tackle the growing genetics waiting lists, with a particular emphasis on doubling the number of rare disease genetic consultants (increase from 4 to 8) and genetic counsellors (increase from 4.5 to 9) as a priority over the coming 2 years.
- We additionally call for the development of a national strategy for genetic and genomic medicine services that reflects best international practice, as recommended in the 2014 Donnai-Newman report.

[1] NTPF: Outpatient by Specialty as at 19/12/2019 – accessed 16/01/2020 - https://www.ntpf.ie/home/pdf//2019/12/nationalnumbers/out-patient/National02.pdf

[2] NTPF: Outpatient by Specialty as at 18/12/2018 – accessed 16/01/2020 - https://www.ntpf.ie/home/pdf//2018/12/nationalnumbers/out-patient/National02.pdf

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Key ask 3 – Legislation to expand New Born Screening

Through the heel-prick test, parents of newborns in Ireland are offered the opportunity to have their baby tested for 8 different rare conditions shortly after birth. The number of diseases tested for in other countries is significantly higher than the Irish figure. With the potential for early diagnosis and treatment, and the associated improved outcomes and long-term care savings, we must expand our newborn screening panel to, at minimum, bring us in line with best-practice across Europe (40 different rare diseases). We have an opportunity to be best-in-class and should strive to be a leader in this field of medical science.

- We are calling for legislation to be brought forward and enacted in the next Dáil to ensure that we meet and exceed EU standards for New Born Screening.

About Rare Diseases

A rare disease is a disease that affects fewer than 5 in every 10,000 people. There are currently 8,000 described rare diseases. Rare diseases are characterised by a wide diversity of symptoms and signs that vary not only from disease to disease but also from patient to patient suffering from the same disease. Rare Diseases are associated with multiple impairments including cognitive, developmental, intellectual, mental, physical and sensory, or some combination of these symptoms. Rare diseases are chronic, progressive, degenerative and often life-threatening.

About the Rare Disease Taskforce

The Rare Disease Taskforce was established in 2013 by Health Research Charities Ireland (HRCI www.hrci.ie; then the Medical Research Charities Group (MRCG)), the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI www.ipposi.ie) and Rare Diseases Ireland (RDI www.rdi.ie; then the Genetic and Rare Disorders Organisation (GRDO)). The Taskforce brought the three umbrella organisations together to work collaboratively to ensure that the needs of the Rare Diseases stakeholders were represented in development and implementation of the National Rare Disease Plan for Ireland 2014 – 2018.

The Rare Disease Taskforce draws its membership from a wide range of stakeholders, including the umbrella groups, rare disease patient organisations, individuals affected by rare diseases, healthcare providers and industry.