



RARE DISEASE BRIEFING

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GENETICS IS NOT JUST RESEARCH....

- **Genetics in research and innovation**

Advances in research into the genetic course of disease has lead to new hope for therapeutic development

- **Genetic therapies are not a pipe dream**

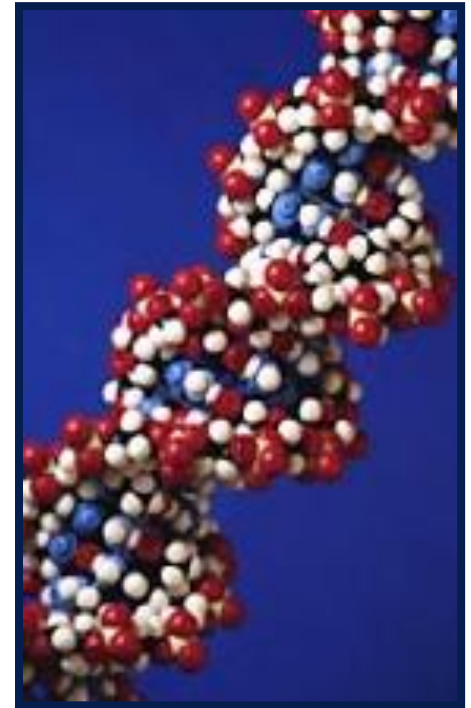
Research laboratories often funded by patient groups are moving to a translational phase

- **Genetic therapies are a reality**

Luxturna for a rare form of childhood blindness approved by FDA in USA and received CHMP approval on Friday September 21, 2018

- **Genetic Therapies herald new era of healthcare delivery**

Personalised medicines, including gene therapies, will target disease revolutionising delivery of treatments and cures for unmet needs



WHAT DOES A GENETIC DIAGNOSIS MEAN AND WHY IS IT IMPORTANT

- 300,000 Irish people will be affected by a rare disease in their lifetime - 80% of rare diseases are genetic in origin.
- One of the largest health constituencies in Ireland yet it remains one of the most marginalised and least served.
- To truly understand the course of genetic diseases, ranging from life limiting disease to physical and/or sensory disease, often with mental and physical developmental delays, a genetic test is required.

STATE OF PLAY....

In Ireland today...

Waiting lists for a diagnosis are increasing year on year

- Currently over 2,800 people on waiting list
 - An increase of 400 from 2017
- 500 people are waiting more than 18 months
 - An increase of 66% compared to 2017

GENETIC SERVICES

In Ireland today...

Because of a lack of investment...

- 4 WTE Consultant Clinical Geneticists
- 6.2 WTE Genetic Counsellors

A counsellor provides diagnosis and impact on prognosis of disease, they need to meet not only affected person but also with all members of a family.

21 PEOPLE AND MORE KEY RELATIVES HAVE DIED WHILE ON WAITING LISTS!

Would this be acceptable
on any other list?



HOW WE CARE FOR OUR MOST VULNERABLE DEFINES THE VALUES OF A COUNTRY

- Are we a progressive country?
- Are we innovative - or is it only those that can shout the loudest that are listened to?
- Caring for a person with a lifelong genetic disease is all consuming - we are here today to represent those who don't have a voice
- We have no catchy slogans or funding for advertising campaigns, just us representing the thousands who want to work with you to develop and implement policies that will leave no one behind

SO ARE WE READY FOR THE FUTURE?

....BECAUSE THE FUTURE IS HERE!

