What Does Success Look Like?

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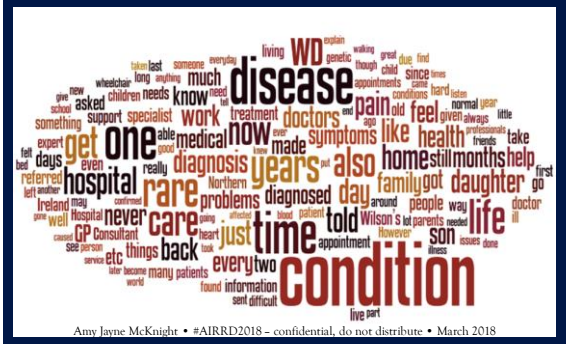
Success Means Different Things…

The Centre of Excellence for Public Health Northern Ireland

We Asked What People Wanted…

More responses in NI than Eng, Scot, Wales



Some Background

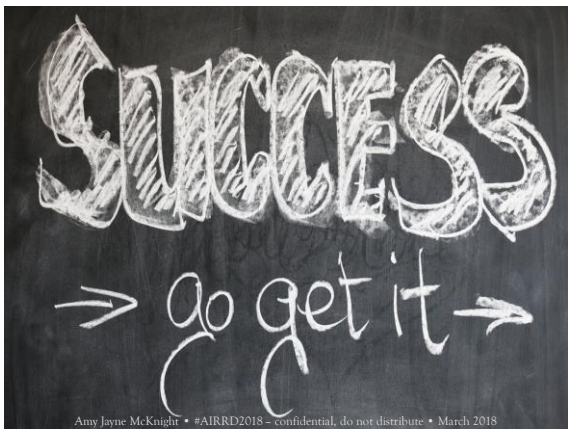
2009: EU Directive - Member States must have a rare disease plan / strategy by 2013.

2013: UK Strategy for Rare Disease published.

Northern Ireland Rare Disease Stakeholder Forum (then including Department of Health and Social Services, HSCB, PHA, clinicians, NIRDP, researchers) developed a statement of intent for Rare Diseases in NI.

2015: launch of NI Rare Disease Implementation Plan.

2017: EU Report



A Vision for a Centre of Expertise for Rare Diseases / Genomic Medicine Centre NI

[We launched the NI GMC in Oct 2015, 100 KGP ongoing – CERDNI in the future?]

Specifically helping with:

- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Co-ordination of care
- The role of research
- Cross-border partnerships

NI has unique potential to develop a rare disease registry and information hub for patients and healthcare professionals. Local expertise and experience in epidemiology, statistics, informatics, rare disease genetics, data management, registries, existing resources, NIECR, NIADRC, and data protection are all readily available.

Patient Voices show a Lack of Diagnosis & Information is Critical

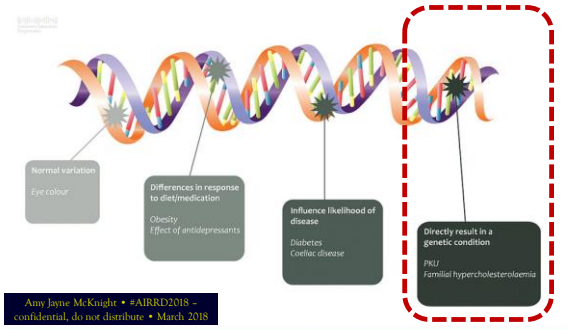
We must Do Better!

Golden Gate Bridge
Took 4 years to build

Lack of Diagnosis

- >5 years average time to diagnosis
- ½ patients received ≥ 1 WRONG diagnosis *before getting the right one*
- ~¼ rare diseases have a molecular cause known: *better tools required.*

Genetic Variation has Many Roles:



Northern Ireland Rare Disease Implementation Plan on Improving Diagnosis [2015]

- ***"Diagnosis"*** – mentioned 29 times in this plan
- **'Diagnosis** and early intervention' one of six key themes
- *'Deliver evidence-based **diagnosis'*** commitment
- *"To establish a **Genomics Medicine Centre** in Northern Ireland and participate in the **100,000 genomes project**".*

We asked for it,
We found the evidence for it
…& **We** got it!
#StrongerTogether

Improving diagnosis…

NI Genomic Medicine Centre launched (2015) whereby 1,278 rare disease people in NI will receive whole genome sequencing (recruitment early 2017 – September 2018)

- ✓ New genetic approach that increases the chance of diagnosis for $^{2}/_{5}$ individuals with a rare disease
- ✓ NI using state-of-the-art 'multi-omic' approaches to improve the speed and accuracy of diagnosis
- ✓ This project may also help explain how a disease will develop and identify the best treatment for patients.

Improving diagnosis…

- ✓ Pilot project that is being evaluated in NI
  - ✓ >200 rare diseases are included
- ✓ Bringing genomics into mainstream medicine
  - ✓ 12 medical specialities on board for pilot
- ✓ Not everyone will get a diagnosis or a particular treatment based on their results
- ✓ **We are still learning!**
- ✓ Major benefit of 100 KGP and NIGMC is to help local individuals living and working with rare diseases
- ✓ Helping train local folk and build capacity in these new approaches + develop new information tools

First patients diagnosed through the 100,000 Genomes Project

Posted on March 11, 2015 at 10:21 am

The first family are set to benefit from ground-breaking developments in whole genome sequencing. Health Secretary Jeremy Hunt will today (Wednesday 11 March) meet the first patients to be diagnosed with rare disease through the 100,000 Genomes Project. They will now receive effective, personalised treatment, as well as helping prevent future generations who share their DNA from suffering a life of uncertainty about similar symptoms.

Newcastle University and Hospitals worked with Genomics England to analyse the genomes and validate the findings as part of the project's pilot scheme.

Now receive effective, personalised treatment

+

Help prevent future generations who share their DNA from suffering a life of uncertainty about similar symptoms

First children receive diagnoses through 100,000 Genomes Project

Posted on January 11, 2016 at 6:09 pm

The first children to receive a genetic diagnosis through the 100,000 Genomes Project have been given their results at Great Ormond Street Hospital (GOSH), part of the North Thames NHS Genomic Medicine Centre.

Both Georgia Walburn-Green and Jessica Wright had rare, undiagnosed, genetic conditions when they joined the Project. Whole genome sequencing pinpointed the underlying genetic changes responsible for their conditions.

Mum said *"As soon as we were on the project I felt a huge sense of relief".*

Georgia now has a molecular diagnosis

By 2019, **1,278 whole genomes** will be sequenced from individuals living in NI

Getting the right diagnosis:

- Empowers people living and working with rare diseases
- Provides relevant information
- Facilitates the best treatment
- Enables connections with disease focused support groups / peers / experts
- Allows effective planning for the future

NI contributes to the 100,000 Genomes Project

www.genomicsengland.co.uk/50000-genomes-landmark/

20 UK leads the world as 100,000 Genomes Project hits the 50,000 genomes landmark to transform NHS patient care

What Next?

Northern bald ibis

Believed extinct until rediscovered in 2002

Lack of Information

- >80% individuals report accessing relevant information is difficult: *>70% of GPs struggle to identify / manage rare diseases*
- >½ patients have >10 medical appointments each year: *no clear care pathway / patient journey / linked systems*
- ½ of rare diseases do not have a specific support group
- Patients, families, carers often become **'Expert Patients'**: *patients at the centre of their care.*

Information Priorities for NI

1. **Dedicated NI Rare Disease Website / Information Hub for Rare Diseases**
2. **Centralised NI Registry / Registries for Rare Disease**

- NI Rare Disease Coordinator to Facilitate Access to Information
- Specialised Rare Disease Training for Professionals
- Communication and strong inter-professional networks (MDTs)
- Online training modules; generic and specific
- Access to Good Disease Specific Information

Thank You!

To many people to count…everyone who contributed, in particular Julie McCarroll, **Cheryl Flanaghan,** Tara Moore, Andrew Nesbitt, Helen McAneney, Julie McMullan, Ashleen Crowe, Katie Kerr, Jill Kilner Fiona Stewart & Shane McKee with BHSCT team delivering 100 KGP …

- Partnership is essential:

- ✓ Co – develop
- ✓ Co – produce
- ✓ Co – enact
- ✓ Co – maintain / review / improve

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To Future Progress