DO YOU NEED A PATIENT REGISTRY?

Mary Bythell, Head of Rare Disease Registration, Public Health England
Jeanette Anderson, Rare Disease Data Liaison, Public Health England
Angela Stringer, Duchenne Muscular Dystrophy Register Curator, Action Duchenne
Rebecca Cosgriff, Registry Lead, Cystic Fibrosis Trust
National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)

What we do and how we can work together
Mary Bythell
Head of Rare Disease Registration

Kay Randall
Thames Valley Regional Lead & National Clinical Training Lead

Jeanette Aston
Rare Disease Data Liaison

Adenike Adesanya
Registration Officer
Genetic Alliance Annual Conference

• Introduction to NCARDRS
• Congenital anomalies
• Rare disease data collection expansion
• Working together
NCARDRS – key deliverables

- **support and empower patients** and their carers, by providing a national register of their disease or disorder.
- provide a **resource for clinicians** to support high quality clinical practice.
- provide **epidemiology and monitoring** of the frequency, nature, cause and outcomes of these disorders.
- **support research** into congenital anomalies and rare diseases.
- **inform the planning and commissioning** of public health and health and social-care provision.
- Provide a resource to **monitor, evaluate and audit** health and social-care services, including the efficacy and outcomes of screening programmes.
Information governance

• NCARDRS has Section 251 approval to collect non-consented patient identifiable data

• Data sharing agreements (DSAs) with most relevant trusts

• NCARDRS has legal bases under GDPR to process both personally identifiable data and special category data

• Patients have a right to opt out
Fair processing

National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)

If you, your unborn baby or child has a suspected or confirmed rare disease or congenital anomaly, clinicians will pass your information onto NCARDRS.

What are congenital anomalies?
Congenital anomalies are structural defects or abnormalities of an organ or body structure present at birth. They range from minor to major and can affect development in different ways.

What are rare diseases?
Rare diseases are conditions that affect a small number of people compared to the general population and, because they are rare, can be difficult to diagnose, treat and/or prevent. A disease is considered rare when it affects 1 person in 2,000 or fewer.

Why we collect this information?
- To provide patients and carers with information relevant to a disease or condition
- To give health professionals information to monitor and improve clinical practice
- To help patients and people with rare diseases
- To monitor and evaluate antenatal and newborn screening programmes.

www.gov.uk/ncards

The information held on the register is strictly confidential and held in accordance with the Data Protection Act.

If you have any questions about NCARDRS, please discuss them with your midwife or doctor, who can also provide you with our patient information booklet.

Public Health England, 130-135 Waterloo Road, Wellington House, London, SE1 9DS.
Tel: 030 7664 3000. Email: ncards@phe.gov.uk

Screening tests for you and your baby

Important information about the screening choices you will have during and after your pregnancy.

Further appointments: You will be contacted sooner if there is thought to be any problem with your baby.

What happens to my baby's blood spot card and data after screening?

After screening, blood spot cards are stored for at least 5 years and may be used:
- to check the result or for other tests recommended by your doctor
- to improve the screening programme
- for research to help improve the health of babies and their families in the UK.

This research will not identify your baby, and you will not be contacted. The use of these blood spots is governed by the code of practice available from your midwife, or on this website.

If it looks like your baby has sickle cell disease or thalassaemia, we will pass the information to the National Congenital Anomaly and Rare Disease Registration Service.
Public Health England – what we cover

Cancer
NCRAS

Rare disease

Congenital anomalies
NCARDRS

Infectious disease
Health protection
Public Health England – what we cover

- Rare disease
- Congenital anomalies
THE NATIONAL CONGENITAL AND RARE DISEASE REGISTRATION SERVICE

- 940 Different congenital anomalies and rare diseases collected
- 244 Data collected from 244 healthcare providers in England
- 42 members of staff working at 8 regional offices
- 30+ years’ information for some regions
- 1 national data management system
- 1st Jan 1985
WORLD CLASS DISEASE SURVEILLANCE: SUPPORTING EVALUATION AND MONITORING OF NATIONAL SCREENING PROGRAMMES

Working with over 40 disease registers worldwide

Meeting European data quality standards

136 Data on antenatal detection rates reported back to all 136 maternity units annually

CONGENITAL ANOMALIES
49% coverage of all births in 2015. 100% coverage today

18 Data feeds from 18 cytogenetic laboratories
Rare diseases data collection

- Enormous task
- Limited resources
- Pragmatic approach – based on specificity, quality & availability and patient benefit
- Not mandatory
- Project-based approach for disease/disease groups
  - Internal
  - Externally funded
Current project examples

- Increasing granularity of congenital anomaly (CA) coding
- Improving post-neonatal reporting of CAs
- Wilson’s Disease proof of concept (with BASL)
- ANCA-associated vasculitis (externally funded)
- Newborn blood spot IMDs – retrospective & prospective
- Piloting molecular genetics test results for specific conditions
Working with NCARDRS

PHE currently supports such data access in two ways:

• Formal requests for release of data
• External funding or sponsorship for staff
Release of data

- Data releases follow the Public Health England Office for Data Release (ODR) process
- Further information is available on the ODR website (see handout)
<table>
<thead>
<tr>
<th>Applicant's organisation type</th>
<th>Number of releases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Academic Institution</td>
<td>49</td>
</tr>
<tr>
<td>Commercial</td>
<td>2</td>
</tr>
<tr>
<td>TraceMedia Projects</td>
<td>1</td>
</tr>
<tr>
<td>UK Biobank</td>
<td>1</td>
</tr>
<tr>
<td>CQC Registered Health or/and Social Care provider</td>
<td>28</td>
</tr>
<tr>
<td>Government Agency (Health and Adult Social Care)</td>
<td>16</td>
</tr>
<tr>
<td>Independent Sector Organisation</td>
<td>37</td>
</tr>
<tr>
<td>Local authority</td>
<td>1</td>
</tr>
<tr>
<td>Other</td>
<td>1</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>134</strong></td>
</tr>
</tbody>
</table>
External funding for staff

- Work must align with PHE’s overall objectives: “Improve and protect the nation’s health”
- Benefits should be mutually beneficial

Two types of employment:
- Through PHE
- Partnership organisation with honorary contract
External funding sources

- NHS England
- Cancer Research UK
- Macmillan Cancer Support
- Teenage Cancer Trust
- Association of British Pharmaceutical Industry (ABPI)
- London Cancer Alliance
- Health Data Insight CiC
- Royal College of Physicians
- Royal College of Surgeons
- Transforming Cancer Services London
- various PhD students or Clinical Fellows from academic partners
Key messages

• Section 251
• Large datasets with linkage permissions
• Project-based approach
• Good clinical engagement
• Enormous potential
• Happy to work with external funders
Thank you

www.gov.uk/guidance/the-national-congenital-anomaly-and-rare-disease-registration-service-ncardrs
Action Duchenne

UK DUCHENNE MUSCULAR DYSTROPHY (DMD) REGISTRY

GENETIC ALLIANCE UK CONFERENCE

25th September 2018
Action Duchenne

- Charity set up in 2001
- By parents refusing to accept ‘no hope’
- Raising funds for support and research
- Registry to find patients for clinical trials
- Engaging with all stakeholders – parents, patients, clinicians, geneticists, researchers and pharma
Action Duchenne

- **UK DMD REGISTRY**
  - Set up in 2007
  - One of the first for DMD
  - Received a grant
  - Parent with IT knowledge
  - Steering committee – clinicians, geneticists, families
  - Support of the DMD community and endorsed by professionals
Action Duchenne

- Voluntary and anonymous
- No 3rd party access
- Progressed from paper to online registration
- Password security and easy access
- Streamlined data collection helps speed up the process to identify patients
- Gene mutation validated by geneticist
- Regulated by Data Protection and GDPR
Action Duchenne

- Data collection:
  - For Duchenne and Becker
  - By parent or participant over 18 years and carriers
  - Contact details
  - Medical – gender, dob, hospital,
  - Genetic – disease in family, DNA test lab
  - Interventions – Steroid use, surgical procedures, cardiac, bone density score
Action Duchenne

– Functionality – ventilation, motor function, wheelchair use, behavioural or learning difficulties.
– Trials
– Mediation – supplements, equipment
– Agreement to Terms and Conditions
– Consent form to be signed
– Access for patients at all times and a patient report
Action Duchenne

• What to collect?
  – Collecting relevant data
  – Steering Committee with specialist knowledge
  – Part of the TREAT-NMD Global Registry

  – WHY?
  – Record prevalence and natural history of the disease
  – Future research
  – Assist acceleration of research programmes and contact between health professionals, researchers and patients
Action Duchenne

• Registry Curator
  – Personal contact
  – Helps complete the process
  – Responds to external enquiries
  – Contact with other international registries through TREAT-NMD
Action Duchenne

• THANK YOU

• Any questions?
Cystic Fibrosis our focus

The benefits of a patient registry

Rebecca Cosgriff, Director of Data & Quality Improvement
Cystic Fibrosis

What, exactly?
Annual Reviews

9,887 annual reviews recorded

That’s 192 more than 2016

Population age

20 is the median age of people with CF on the Registry

60.6% of people on the Registry are aged 16 and over.
UK CF Registry

A very short introduction
“The future is already here, it’s just not very evenly distributed.”
Cystic Fibrosis

our focus

Thank you

cysticfibrosis.org.uk
WORKSHOP ROOMS

Managing online communities
Action Room 2 (off auditorium)

Building a strong case for support
Conference Room (Mezzanine level)

Working with Media
Auditorium

Do you need a patient registry?
Garfield Weston Action Room (off auditorium)

Producing patient information and leaflets
Annex Room (Lower Ground floor)