The National Congenital Anomaly and Rare Disease Registration Service

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Current congenital anomaly registration across England

- Only 49% of births are currently monitored by 7 regional registers.
- Currently inequitable registration and surveillance of congenital anomalies across the population.
- Unable to provide comprehensive outcome data for screening programmes in England
Public Health England’s mandate

- Chief Medical Officer’s 2011 annual report:
  
  “Public Health England must ensure nationwide coverage of the congenital anomaly register.”
Expanding coverage of congenital anomaly registration across England

- Creating 5 new regional teams to cover:
  - North West
  - East of England
  - South East
  - London
  - Yorkshire and Humber
- In National Cancer Registration Service (NCRS) offices:
  - IG framework in place to collect patient identifiable data
Establishing a federated model

Central team:
- Programme Manager
- IT development
- Administrative support
- IG compliance
- Dataset liaison

Regional registration team
Opportunities

75% of rare diseases identified at birth or shortly after

Data on the remaining 25% rare diseases identified in adulthood

A comprehensive national register for rare diseases in England
Our vision

To develop and run a cost effective national congenital anomaly and rare disease registration and surveillance service for England.

as a resource for

patients, families and carers, researchers, public health, safety, audit, and equity monitoring.
Working within the UK and European context

- Close links with genomics
- Commitments within the UK Strategy for RD
- European and International interoperability
National data system and data collection

- Single data system developed to provide:
  - a high degree of electronic data capture
  - timely data collection
  - local and national data feeds
  - improved quality assurance
  - timely feedback to clinical teams

- System and collection based on NCRS model
Maternity Minimum Dataset

Clinical Data from Fetal Medicine Teams

Regional Genetics Laboratories

Biochemistry laboratories

Maternity Minimum Dataset

Notifications from ultrasound

Antenatal Screening Programmes

Antenatal Care

Birth

Postnatal

Childhood

Later onset

Congenital Anomalies

Rare Diseases

Hospital Episode Data

Clinical Data from Neonatal and Paediatric Teams

Notifications of miscarriage

National Termination of Pregnancy Data

Existing Individual Rare Disease Registers

Genomics England

Regional Perinatal Pathology

ONS Vital Statistics

Regional Perinatal Overview Panel

Newborn Screening Programmes

Child Death Overview Panel

Regional Clinical Genetics Services

Specific requests to individual GPs

Regional Clinical Genetics Services

Regional Clinical Genetics Services

Patient Portal

Patient/ carer self registration, PROMS, QOL

ONHSE specialised commissioning Registers

Regional Perinatal Epidemiology Unit

Newborn Screening Programmes

MBRRACE-UK

National Perinatal Epidemiology Unit

Regional Perinatal Pathology

Notifications of miscarriage

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High level timescales and key next steps

• March 2015: New regional teams established, migrate existing registration staff, new data management system in place
• April 2015: National CAR operational
• December 2015: Rare disease data sharing arrangements in place
• December 2016: Patient portal in place
Fighting congenital anomalies and rare diseases with information

Christine Harvey, Jem Rashbass and Sarah Stevens, 23 July 2014 — A single knowledge and intelligence service, Chief Knowledge Officer, Reducing preventable deaths

We hear a great deal about the common illnesses that affect many people such as heart disease, diabetes and cancer. However, we hear much less on rare diseases. Each