Public Health England (PHE) launched the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) on 1 April 2015. Prior to this, congenital anomaly registries existed in some regions of England covering 49% of the population. In response to the UK Rare Disease Strategy and the Chief Medical Officer’s recommendation and consultation with stakeholders, a national system for congenital anomalies and rare disease in England was established. Data collection for congenital anomalies began in new regions for babies expected from 1 April 2017. Building on the established infrastructure and experience of national congenital anomaly reporting, NCARDRS has undertaken a discovery phase to explore methods of registering non-structural, non-chromosomal rare diseases, including those that present in later childhood and adulthood. Working with clinicians, clinical networks, patient groups, academics and other government agencies, we have begun national reporting of inherited metabolic disease and some rare rheumatic conditions, undertaken extensive validation of national routinely collected data, piloted the use of molecular genetic diagnostic data to support rare disease registration and have built strong multi-disciplinary collaborations to strengthen existing registration efforts and underpin further expansion.

**Information Governance**

NCARDRS has legal permission granted under Section 251 of the NHS Act 2006 to collect data without patient consent and we work within strict information governance guidelines. We have permission from the Confidentiality Advisory Group of the Health Research Authority to collect information on all suspected and confirmed cases of congenital anomaly and rare disease in England as defined by EUROCAT and Orphanet. Patients have an absolute right to opt-out.

**National Coverage for Congenital Anomalies in 2017**

The first full year of congenital anomaly data was available nationally for England as a whole for those born in 2017. National prevalence was 208 cases per 10,000 total births and ranged regionally from 169.9 in regions reporting their first year of data, to 262.2 for regions with existing registration.

Many congenital anomalies are also rare diseases and so the experience of extending geographical coverage of the existing regional registers to achieve national ascertainment has significantly informed rare disease data collection and output generation.

**Rare Disease Expansion – areas of focus**

- Molecular diagnostic data
  - Pilot phase
  - Renegotiating data feeds
- Later onset conditions
  - Rare rheumatic disease
  - Wilson’s Disease
  - Early onset diabetes conditions
- Collaborations
  - Academics
  - Patient groups
  - Clinicians and clinical networks
  - Consented & other studies
  - Other government agencies
- Accessible datasets
  - Hospital admissions data
  - Mortality data
  - 1st care prescribing data
  - Cancer registry
  - Other national datasets


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**INTRODUCTION**

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**NCARDRS remote access to electronic patient records**

**Hospital clinicians**

**Positive predictive values**

- Microscopic polyangiitis 98.4% (91.3-100.0%)
- Takayasu's arteritis 91.7% (81.5-99.8%)