

NCARDRS: A population-based congenital anomaly and rare disease register for England.

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INTRODUCTION

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.

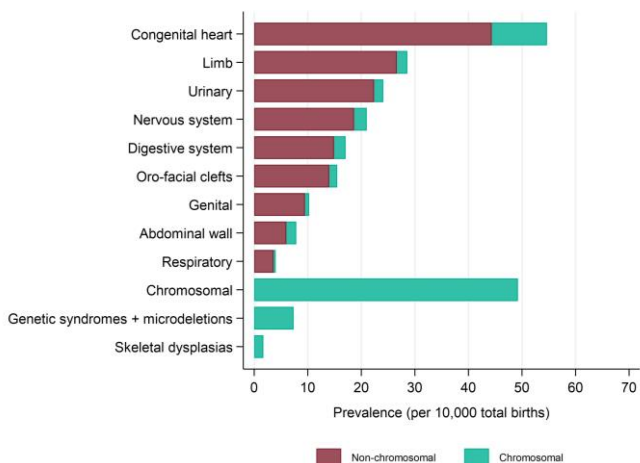
Information Governance

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 2018

The first full year of congenital anomaly data was available nationally for England as a whole for those born in 2018. 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.

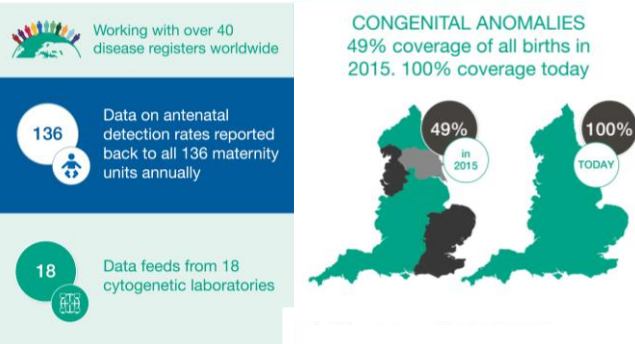
Many congenital anomalies are also rare diseases and so the experience of extending geographical coverage of the existing regional registers to achieve national ascertainment has significantly informed rare disease data collection and output generation.



National prevalence by anomaly group for births in 2018 for England



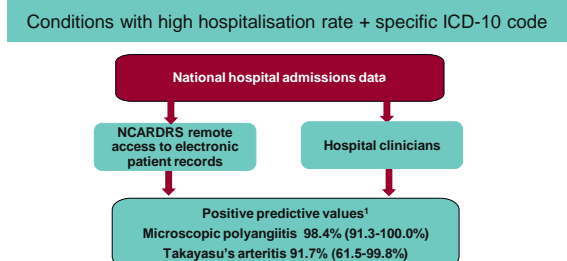
WORLD CLASS DISEASE SURVEILLANCE: SUPPORTING EVALUATION AND MONITORING OF NATIONAL SCREENING PROGRAMMES



Rare Disease Expansion – areas of focus

- Congenital anomalies**
 - National coverage
 - Increased granular coding
 - Improved post-natal diagnosis ascertainment
- Molecular diagnostic data**
 - Pilot phase
 - Renegotiating data feeds
- Later onset conditions**
 - Rare rheumatic disease
 - Wilson's Disease
 - Early onset diabetes conditions
- Metabolic disease**
 - Newborn blood spot
 - Specialised services
- Collaborations**
 - Academics
 - Patient groups
 - Clinicians & clinical networks
 - Consented & other studies
 - Other government agencies
- Accessible datasets**
 - Hospital admissions data
 - Mortality data
 - 1st care prescribing data
 - Cancer registry
 - Other national datasets

Rare Disease Expansion – example of validating routinely collected data



National data collection and analysis for rare diseases and congenital anomalies is a powerful basis from which to understand these conditions. Using a multi-source approach to data collection enables us to triangulate, confirm and complete cases and mitigates limitations of individual data sources. Working closely with stakeholders is key to high-quality national congenital anomaly and rare disease registration.

*Pearce FA, Rutter M, Griffiths B, et al. O36 Validation of methods to enable national registration for rare autoimmune rheumatic diseases. Rheumatology 2020;59. doi:10.1093/rheumatology/keaa110.035

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