National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) Workplan 2021-24

This plan sets out NCARDRS' priorities for the period 2021-24 within the context of the National Disease Registration Service's (NDRS) vision, building on what we have achieved since our launch in April 2015.

Background

NCARDRS exists to develop and run a comprehensive population-based registration service that collects and quality-assures data on congenital anomalies and rare diseases across the whole population in England.

Disease registration is key to intelligent public health and healthcare delivery. NCARDRS sits within the NDRS, alongside the National Cancer Registration and Analysis Service (NCRAS). We provide expert analysis and interpretation of the data we collect, which is used as a source of intelligence for a range of stakeholders.

NCARDRS is a much needed resource to support individual patients, their families, clinicians, research, service delivery, healthcare performance, commissioning and public health. We:

- 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, rare diseases and precision medicine including basic science, cause, prevention, diagnoses, treatment and management through access to existing data and collaboration on new data streams
- 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.

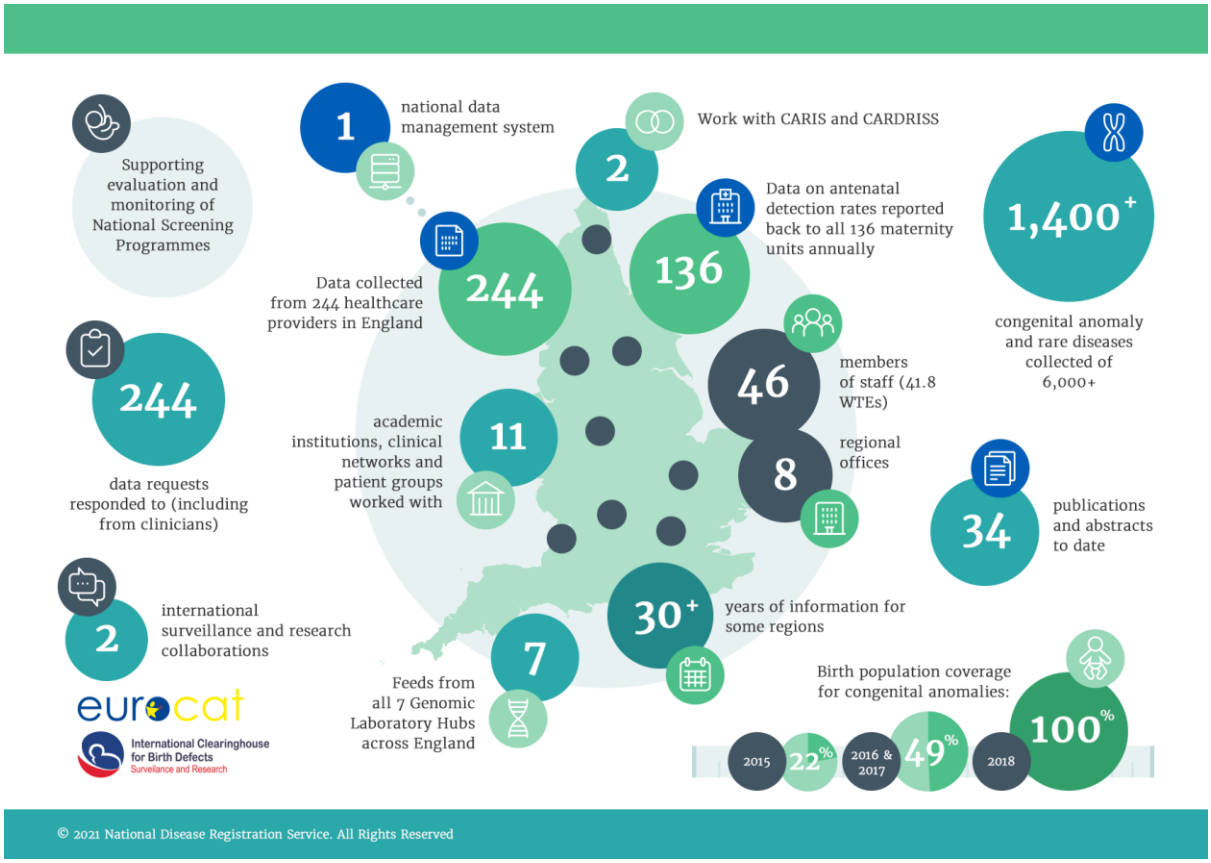
Our approach

Engagement with key stakeholders has been fundamental to delivering our achievements to date. Patients and their families lie at the heart of our work. We work with the Patient Empowerment Group of the Rare Disease UK campaign, managed by Genetic Alliance UK. Clinical champions work with us to ensure the robustness and relevance of our processes and output. Extensive engagement with clinicians, patient groups and other stakeholders have informed the priorities and the overall development of the service outlined within this plan. We will further focus our engagement on working with Genomics England, NHS England, NHS Digital and the Office for Life Sciences to inform and support policy initiatives and ensure alignment with the roll out of the Genomic Medicine Service (GMS). NCARDRS will also play a key role in the delivery and support of the UK Rare Disease Framework, working closely with the DHSC and other partners.

We have built positive working relationships with our data providers to improve data quality and completeness. We continue to review our data collection processes to ensure the burden on clinical staff is kept to a minimum, and are refining our tools and outputs to improve timely feedback to clinical teams. We continue to engage existing and potential data providers to allow us to understand the challenges to data sharing and work together to overcome them, seeking to capitalise on bulk data feeds that can be automated with a minimum of manual extraction for all parties without compromising data quality. These data liaison functions will be crucial to the success of implementing the rare disease data collection expansion and supporting the GMS. For this, we will take a pragmatic approach. We will prioritise collecting data that is currently available while working to identify sustainable systems and processes to collect information on other rare diseases where we have resource to do so.

We have Honorary Contracts in place with leading clinicians and academics in the field. We have established a process with PHE's Office for Data Release (ODR) through which we have managed 224 requests for data during the period 01/01/2015 to 01/12/2020. We have produced four annual congenital anomaly statistics reports, in which coverage of congenital anomalies has expanded from 21% of England in 2015 to full, national coverage in 2018. Our ambitions for the service have led to funded work programmes with partners including the DHSC, the National Screening Programmes and Orphanet.

Our achievements to date: 2015-2020



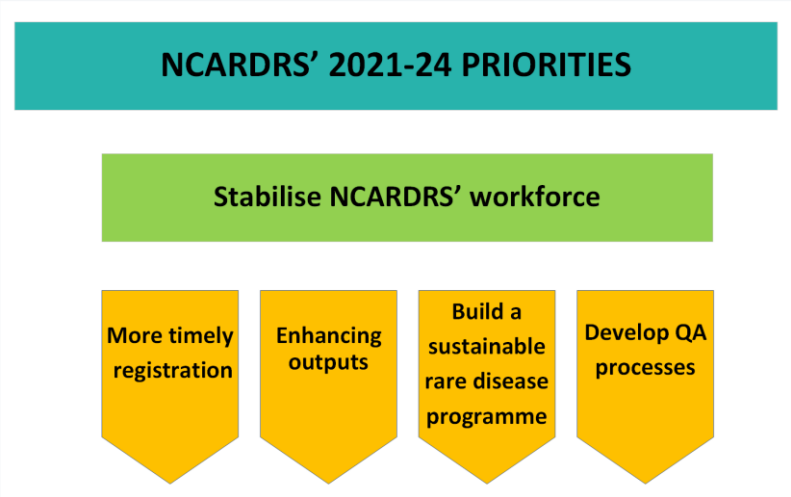
Our focus 2021-2024

High-quality data, and the information derived from it, is central to any healthcare system. We have made good progress in establishing the service over the first five years, with national coverage of over 1,400 congenital anomaly or rare disease data sets collected.

In the period to 2024, NCARDRS will focus on five key priority areas. Central to these is strengthening the service's foundations by stabilising the workforce. NCARDRS has been ambitious in its goals and outputs, and we now need to develop the functions within the service to meet these commitments in a sustainable way for the future. NCARDRS has developed strong collaborations with stakeholders since 2015, securing funding to develop work programmes. This has led to a growing number of short-term funded contracts which we want to sustain to ensure our business-as-usual capacity is appropriately resourced.

Whilst the Covid-19 pandemic highlighted our ability to rapidly generate answers to key clinical questions from within the largest rare disease population in Europe, it also highlighted the need for NCARDRS to position itself more strongly with external stakeholders and patients by becoming more responsive to clinical queries. We will achieve this by automating more registration processes to improve the timeliness of registration, and enhancing routine surveillance to improve understanding of congenital anomaly and rare disease prevalence, pathways and outcomes. NCARDRS is committed to the continuous improvement of data quality, and we will develop our quality assurance processes and continue to drive the development of national coding and classification guidance.

NCARDRS will build on the work from 2018 to expand its rare disease data sets by strengthening and expanding partnership work with stakeholders, including patients, clinicians, academia and the private sector, to deliver proactive and reactive, high-impact and novel output. We will develop automated systems that can deliver sustainable, high ascertainment, prospective reporting.



The deliverables behind each of these priorities are outlined in the following table, in order of importance. They will be labour intensive. The workforce resources that are available to NCARDRS will directly affect the extent of achievement of these aims and the pace of these achievements.

Our priorities

Priorities and deliverables		Lead	Timeframe	Funding source, where external
Priority 1: Stabilising NCARDRS' workforce, securing funding and partnership collaborations **CH** We will ensure that NCARDRS is appropriately resourced, on a sustainable basis, to fulfil business-as-usual commitments. NCARDRS will extend and strengthen its work programmes across analysis and rare diseases by developing partnerships with external stakeholders. Funding opportunities will be sought to sustain programmes of development work, and to maintain integral collaborations for patient and health system benefit.				
Deliverables				
1.1	Scope capacity needed to deliver NCARDRS' business-as-usual work, produce a gap analysis and work with the NDRS Strategic Lead to secure sustainable funding and resource to stabilise NCARDRS' workforce structure.	JS/DL/JR	September 2022	
1.2	Review and secure sustainable asset funding for significant enhancements to CARA.	CH	March 2024	Capital or similar
1.3	Develop a sustainable, longer-term funding model for the rare disease programme, which provides sufficient permanent capacity for the team to deliver expanded project work.	SS	December 2021	
1.4	Strengthen the analytical capacity allocated to NCARDRS to enable enhanced delivery of outputs as new programmes come on stream.	JB/SS	Ongoing	
1.5	Continue to strengthen work with Antenatal and Newborn Screening programmes through ongoing reviews and identifying new areas for collaborations.	SS/MB/CH/ NM/JB	Ongoing to March 2024	Screening
1.6	Review NCARDRS' commitments under the Orphanet collaboration and funding grants.	SS/CH	December 2023	Orphanet
1.7	Engage with the Patient Empowerment Group and other patient stakeholders, to ensure that output is relevant for patients, including focus group discussions, surveys, patient input into projects and publishing a guide to working with NCARDRS.	MB/JB	Ongoing to March 2024	
1.8	Formalise existing, and create new, partnerships to underpin themed programmes of rare disease work, including liver disease and dermatology.	HM	Ongoing to March 2024	Partnership

Priorities and deliverables		Lead	Timeframe	Funding source, where external
1.9	Collaboratively develop a partnership framework for working with a wide variety of partner types across the system, including the NHS, third sector, academia, industry and others, which reflects the needs and priorities of people living with a rare disease or congenital anomaly and NDRS.	HM	September 2022	
Priority 2: Moving to more timely registration and automating registration processes NCARDRS will position itself more strongly with external stakeholders and patients by becoming more responsive to clinical queries. We will achieve this through a higher degree of automated processes within the registration function, providing us with more timely data to utilise and report on.		**JS/DL**		
Deliverables				
2.1	Accelerate national FASP cohort data completion by 1 month to July 2021 for EDD 2019/20, and by a further 3 months to February 2022 for EDD 2020/21, to enable timely reporting of the FASP audit.	JS/DL	April 2022	Capital
2.2	Pilot, refine and implement (close to real-time) registration of notified 18^{+0} to 20^{+6} week screening scans and fetal medicine referral scans, to facilitate NCARDRS' audit of FASP Standard S08 *Referral: time to intervention*. Initiate a pilot registration and audit process by April 2021, and implement final registration and audit process by April 2022 as part of FASP's revised standards.	NM	April 2022	Screening
2.3	Develop maternal NHS waterfalls (biochemistry, HES) and top-up process to achieve near-complete maternal ethnic group capture in FASP cohort 2020/21.	JS/DL	February 2022	
2.4	Complete outstanding validation and development for HES, Viewpoint, Biochem, PDFs and other feeds which we acquire.	JS/DL	March 2024	Capital
2.5	Accelerate national birth cohort data completion by 3 months to December 2021 for birth cohorts 2018, 2019, 2020 and aim to complete cohort data 9-12 months following date of delivery subsequently, to enable timely reporting and surveillance of the data.	JS/DL	December 2021	Capital
2.6	Create a filing system for source data in CARA.	JS/DL	December 2023	
2.7	Create a Data Entry Tool to capture data items not available via the Maternity or Neo-natal national data sets from Trusts and other providers. Embed a means of exchanging patient data (for QA, etc) within the tool to reduce IG risks (ie, a portal).	JS/DL	December 2022	

Priorities and deliverables		Lead	Timeframe	Funding source, where external
2.8	Create a centralised data loading function for the service. Build a structured, automated process as an interim measure whilst longer-term development of the Data Entry Tool and portal/data exchange is completed.	JS/DL	December 2023	
2.9	Move the data liaison function from NCARDRS registration staff to the NDRS data liaison work programme. Put additional resource in place initially to complete Data Security Agreements and agree remote access with Trusts.	JS/DL	December 2022	
2.10	Establish a Dataset Standard to improve data completion. Prepare an application for a Dataset Standard with the Data Standards Board (NHSD) to cover post-natal data for the unique NCARDRS fields and to enable data collection through the Maternity Services and the Neo-natal Services national data collections.	AM	December 2024	
Priority 3: Enhance outputs **JB** We will position NCARDRS as the natural collaborator for congenital anomaly and rare disease projects by using our data to perform routine surveillance to enhance and improve understanding of congenital anomaly and rare disease prevalence, treatments and pathways, and outcomes. NCARDRS will raise its data quality and its profile by producing peer-reviewed publications and reports.				
Deliverables				
3.1	Develop the analysis server to accommodate enhanced analytical requirements, heightened IG controls, support more flexible reporting, and allow routine data linkage within the analysis server to appropriate datasets.	JB	December 2023	Capital
3.2	Evaluate the impact of Covid-19 on the prevalence, outcomes and care pathways for selected congenital anomaly groups and rare diseases.	JB	December 2022	
3.3	Expand reporting for congenital anomalies and rare diseases through peer-reviewed publications and reports	JB	Ongoing to March 2024	
3.4	Produce Screening outputs: continue NCARDRS reporting for the FASP programme, and proceed with expansion to NIPT reporting when the NIPT is rolled out.	JB	Ongoing, annually	Screening
3.5	Produce annual submissions to international bodies: ICBDSR and EUROCAT	JB	Ongoing, annually	
3.6	Produce annual PHE reports on congenital anomalies and rare diseases.	JB	Ongoing, annually	

Priorities and deliverables		Lead	Timeframe	Funding source, where external
3.7	Incorporate NCARDRS data needs into existing NDRS plans to update Cancer Stats, replicating a data reporting tool similar to Cancer Stats to return data to Trusts/Specialised Services (cytogenetic, rheumatology/ inherited metabolic disease).	JB	December 2023	
3.8	Explore badging routine congenital anomaly statistics as national statistics status.	JB	December 2023	
3.9	Develop tools to automate enhanced surveillance of congenital anomalies, including improved reporting at different geographies and routine expansion, and examining effects of deprivation, ethnicity and comorbidities.	JB	December 2023	Capital
3.10	Build on ongoing work with UKTIS, linking maternal prescriptions data, and explore the use of prescriptions data to inform congenital anomaly and rare disease prevalence or risk factors.	JB	December 2023	Capital
3.11	Build collaborations with stakeholders and analytical tools to evaluate environmental exposure, and explore GIS analysis to further understand congenital anomaly prevalence and environmental exposure.	JB	December 2023	Capital
Priority 4: Building a sustainable Rare Disease Programme		**MB**		
NCARDRS will build on the foundations of its rare disease expansion discovery period to strengthen and expand partnership working with rare disease stakeholders, including patients, clinicians, third sector and government agencies, academia and the private sector. Routine rare disease registration will emphasise innovative automated systems that can deliver sustainable, high ascertainment, prospective reporting. Working with our partners, we will establish a programme of project work that delivers proactive and reactive, high-impact and novel output. We will work with the external stakeholders to ensure that data is available to underpin high quality clinical care and research where appropriate.				
Deliverables				
4.1	Working with partners, and across NDRS, publish novel epidemiological studies on selected rare diseases, including cancer outcomes where appropriate.	MB	Ongoing	
4.2	Expand existing NDRS data feeds (Badger, Blueteq, pathology feeds), and explore and secure access to new data sources, with associated CARA development to support a high level of automation for rare disease registration.	MB	March 2024	
4.3	Position NDRS as the obvious collaborator for rare disease studies, becoming the ‘go to’ partner for assessing population healthcare needs according to place, diagnostic and care pathways; treatments; variation in access and outcomes; as well as research.	MB	Ongoing	

Priorities and deliverables		Lead	Timeframe	Funding source, where external
4.4	Work with the Newborn Blood Spot (NBBS) Screening team, and external stakeholders, to support the NBBS Screening programme with the collection of identifiable data, appropriate data sharing and data linkage, and collaborate on outcome assessments.	MB	March 2024	Screening
4.5	Working with patient partners, develop a patient self-reporting system for congenital anomalies and rare diseases that strives to be inclusive and equitable.	MB/JS/DL	September 2023	
4.6	Strengthen coding and classification of rare disease processes: • develop and implement a rare disease diagnostic text library to support automated registration • working across NDRS, prepare for the use of routine SNO-MED CT coded datasets to support rare disease registration utilising primary and secondary care data.	MB	September 2022	
4.7	Work with national organisations, such as GEL and NIHR, to identify funding and establish a joint exemplar project to demonstrate the value of synergistic partnerships through novel output.	MB	March 2022	Partnership
4.8	Build a pseudonymisation function for molecular data to pseudonymise molecular genetic feeds from Genomic Laboratory Hubs. Establish prospective data feeds on rare disease test data from the Genomic Laboratory Hubs.	SH/MB	March 2024	
Priority 5: Developing Quality Assurance processes		**JS/DL**		
NCARDRS is committed to the continuous improvement of data quality and reporting, through agreeing and achieving national quality standards for data processing, analysis and outputs. We will continue to drive the development of national congenital anomaly coding and classification guidance by referencing clinical evidence and drawing on NCARDRS' clinical expertise, as well as through ongoing collaborations with external experts at national and international level.				
Deliverables				
5.1	Achieve nationally agreed processes in the quality assurance of the NCARDRS data, with even ascertainment pre-submission to EUROCAT, and pre-publication of our routine reporting and statistics.	JS/DL	December 2022	
5.2	Work across NDRS to utilise expertise and technical solutions to create an automated system for case-level data quality assurance checks.	JS/DL	April 2022	
5.3	Complete the Coding Tool, and maintain accurate and up-to-date guidance.	JS/DL	December 2022	

Priorities and deliverables		Lead	Timeframe	Funding source, where external
			(maintenance ongoing)	
5.4	Move the Coding Tool out of Access into a 'system', and build this system.	JS/DL	April 2022	

Key to abbreviations

JS	Jill Smith
JB	Jenny Broughan
NM	Nicola Miller
MB	Mary Bythell
CH	Christine Harvey
SS	Sarah Stevens
SH	Steven Hardy
DL	Donna Lloyd
AM	Andy Murphy
HM	Heather McLean
JR	Jane Richardson