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RESEARCH ARTICLE

Detection rates of a national fetal anomaly screening programme: A national cohort study

Nicholas Aldridge[1] | **Pranav Pandya**[2] | **Judith Rankin**[3] | **Nicola Miller**[1] | **Jennifer Broughan**[1] | **Nadia Permalloo**[4] | **Annette McHugh**[5] | **Sarah Stevens**[1]

[1]National Congenital Anomaly and Rare Disease Registration Service, NHS Digital, Leeds, UK

[2]University College London Hospitals NHS Foundation Trust, London, UK

[3]Population Health Sciences Institute, Newcastle University, Newcastle upon Tyne, UK

[4]Screening Quality Assurance Service, NHS England and NHS Improvement, London, UK

[5]NHS Fetal Anomaly Screening Programme, NHS England and NHS Improvement, London, UK

Correspondence
Nicholas Aldridge, National Congenital Anomaly and Rare Disease Registration Service, NHS Digital, UK.
Email: aldridgenicholasi@gmail.com

Abstract

Objective: To measure condition-specific detection rates for 14 physical conditions screened for by the NHS fetal anomaly screening programme (FASP) fetal anomaly (FA) ultrasound scan.

Design: Retrospective audit of 12 694 diagnoses across a 3-year national cohort.

Setting: All English NHS and crown-dependency hospital trusts providing maternity services.

Population: Pregnancies booked for maternity services with an expected date of delivery between 1 April 2017 and 31 March 2020 and at least one diagnosis of a condition screened for by FASP.

Methods: Active multi-source ascertainment, linkage, audit and validation of clinical information to identify the subset of diagnoses meeting the condition-specific positivity threshold for the FA scan.

Main outcome measure: The accuracy of the FA scan compared with diagnostic reference standards.

Results: FA scan detection rates were: anencephaly 96.3% (95% confidence interval [CI] 81.7–99.3%), atrioventricular septal defect: 69.2% (95% CI 65.8–72.4%), bilateral renal agenesis: 98.7% (95% CI 95.4–99.6%), cleft lip: 89.5% (95% CI 87.8–90.9%), congenital diaphragmatic hernia: 60.8% (95% CI 56.5–65%), Edwards syndrome: 73.8% (95% CI 67.5–79.3%), exomphalos: 59.4% (95% CI 49.4–68.7%), gastroschisis: 88.6% (95% CI 79–94.1%), hypoplastic left heart syndrome: 92.7% (95% CI 90–94.8%), lethal skeletal dysplasia: 93.2% (95% CI 88.6–96%), Patau syndrome: 82.3% (95% CI 72.4–89.1%), spina bifida: 93.8% (95% CI 91.8–95.3%), tetralogy of Fallot: 75.4% (95% CI 72.1–78.4%) and transposition of the great arteries: 84.9% (95% CI 81.7–87.5%).

Conclusions: The performance of the FA scan is above the expectations set in 2010 for most conditions. For the remaining conditions, the majority of fetuses and babies affected are detected before the FA scan.

KEYWORDS
antenatal, congenital anomaly, fetal, radiology, screening, sonography, ultrasound

1 | INTRODUCTION

Congenital anomalies are one of the identifiable causes for stillbirth, neonatal mortality and morbidity. Data from 11 European countries gives the birth prevalence of congenital anomalies at 26.9 per 1000 births.[1] On the recommendation of the UK National Screening Committee, in 2010 the National Health Service Fetal Anomaly Screening Programme (FASP) introduced a national protocol[2] for an 18^{+0}–20^{+6}-week gestation fetal anomaly ultrasound scan (henceforth 'FA scan') to screen for the presence of 11 conditions for which the growing but still limited data suggested a detection rate of at least 50%.[3] This made England the first country worldwide to introduce such a standardised approach to ultrasound screening in pregnancy. Later subdivided into 14 conditions, the national guidance includes expected condition-specific detection rates based on a review of 20 years of published literature.[4,5] These range between 50 and 98%[2] and remain unchanged in 2021 (see Table 1 for expected detection rates for the 14 audited conditions).[6]

All screening programmes must weigh benefits against the harms of 'overdiagnosis, overtreatment, false positives, false reassurance, uncertain findings and complications.'[7] Moreover, women have a choice of whether to have the FA scan, and making a personal informed choice requires accurate information on the 'risks, limitations, benefits and uncertainties'[8] of the test. Since 2010, local and regional attempts to audit FA scan detection rates have been of small scale without uniform methodology to ensure comparability, constrained by the low prevalence of the conditions and the challenge of ascertaining diagnoses not made at the time of screening.[9] Measuring FA scan detection rates at the population level is therefore crucial to determine whether FASP is achieving its expected detection rates.

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)[10] was established in 2015 to provide national surveillance of suspected and confirmed congenital anomalies for England, actively ascertaining clinical information from multiple sources across antenatal and postnatal settings. NCARDRS can accurately identify diagnoses following false-negative screening results and developed an approach to auditing FA scan performance at a national level. The aim of this study is to evaluate the detection rates of 14 pre-defined conditions at the FA scan in a national cohort, against FASP expected detection rates.

TABLE 1 Conditions audited and FASP expected detection rates

Condition	FASP expected detection rate (%)
Anencephaly	98
Atrioventricular septal defect	50
Bilateral renal agenesis	84
Cleft lip ± palate	75
Congenital diaphragmatic hernia	60
Edwards syndrome	95
Exomphalos	80
Gastroschisis	98
Hypoplastic left heart syndrome	50
Lethal skeletal dysplasia	60
Patau syndrome	95
Spina bifida	90
Tetralogy of Fallot	50
Transposition of the great arteries	50

2 | METHODS

2.1 | Participant selection

A total of 133 NHS trusts in England offer the FA scan as part of FASP services, and three trusts located in crown dependencies follow comparable FASP protocols (henceforth 'providers'). The study cohort was consecutively sampled from pregnancies booked for maternity services at all providers, with an expected date of delivery between 1 April 2017 to 31 March 2020, where the baby or fetus was diagnosed with one or more of the 14 conditions screened for by the FA scan. Where a baby or fetus had two or more separate diagnoses of conditions screened for by the FA scan, they were counted once for each.

Data were actively ascertained and linked at record level by the national team of disease registration officers of NCARDRS. Data were linked from multiple sources (see Table 2) to maximise, validate and complete the details of each pregnancy and ensure the capture of diagnoses made outside the initial screening setting. Diagnoses were captured after referral, postnatally, following termination of pregnancy for fetal anomaly, by other clinical services or in other parts of England. NCARDRS collected the study data under legal permissions granted under Section 251 of the NHS Act 2006. NCARDRS only collects data where there is suspicion of a congenital anomaly.

2.2 | The index test

The FA scan is a screening test and does not offer diagnostic certainty, but an indication of increased chance of a condition. It consists of the 'base menu' examination detailed in Table S1: a sequence of ultrasound measurements and confirmations of expected development specific to each anatomical structure, and a cardiac protocol requiring five views of the structures of the fetal heart.[11] FASP offers the FA scan to all eligible pregnant women in England who first present to NHS maternity services before 22^{+0} weeks of gestation, or up to 23^{+0} weeks of gestation where the provider's ultrasound capacity can accommodate this.[12] FASP guidance is for the FA scan to be completed between 18^{+0} and 20^{+6} weeks of gestation, with a single recall scan up to 23^{+0} weeks where the result of the first scan was incomplete, although providers can offer the initial scan after 20^{+6} weeks where appropriate.[12]

Where one of the 14 conditions is detected at the FA scan, FASP guidance is to offer further diagnostic testing with

TABLE 2 Data sources

Data source	Level
Biochemistry laboratories feeds	National
Cytogenetics laboratories feeds	National
Cardiac services feeds	Local/ tertiary
Cleft services feeds	Regional
Fetal Medicine systems (Viewpoint/Astraia) feeds	Local
Patient Administration System extracts feeds	Local
Patient Administration System surgical data feed	Regional
BadgerNet neonatal remote access	Local
Maternity Information system remote access	Local
Radiological systems (CRIS) remote access	Local
Fetal anomaly scan reports	Local
Fetal medicine scan reports	Local/ tertiary
First-trimester scan reports	Local
Individual case notifications	Local
Post-mortem reports	Regional
Hospital Episode Statistics Admitted Patient Care access	National
Office for National Statistics Death Registration access	National
NHS Summary Care Record access	National
NHS Demographics Batch Service access	National

specialist services, for example referral to a fetal medicine specialist, fetal echocardiography or invasive prenatal diagnosis. Where there are no unexpected findings at the FA scan and further testing is not undertaken, diagnosis may not be made until later in pregnancy, after delivery, or even multiple years postnatally. These gold standard diagnostic confirmations are the reference standard against which the FA scan was evaluated; they are presented in Table S2 alongside the condition-specific positivity cut-off for the FA scan used in the study. FA scan results were commonly available to the assessors of the reference standard where the scan result provided the indication for further testing, but may not have been available for diagnoses made in other settings. Reference standard results were not available to the performers or readers of the FA scan.

2.3 | Calculations

The aim of the study was to measure detection rates within the context of an existing national screening programme, not to attempt to measure the sensitivity of the index test in a neutral or objective setting. As such, condition-specific FA scan detection rates were calculated as the number of participants for whom the index condition was detected by the FA scan (the positivity threshold was met), divided by the number of participants meeting the diagnostic reference standard for the index condition who were eligible for and did not decline the FA scan. The denominator therefore included participants whose FA scans were incomplete after two attempts, and participants who did not receive a complete FA scan when they were eligible to. Where a baby or fetus had two or more separate diagnoses of conditions screened for by the FA scan, each was audited independently against its index condition-specific positivity threshold and represented separately in the calculations. This allowed the calculations to be sensitive to occasions where FA scan performance varied between conditions for the same baby or fetus.

FASP offers routine ultrasound at 11^{+2}–14^{+1} weeks of gestation to confirm viability, date the pregnancy and diagnose multiple pregnancies with the option of the Combined Test to screen for Down, Edwards and Patau syndromes. Women presenting after this gestational window are offered a dating scan and the Quadruple Test to screen for Down syndrome only at 14^{+2}–20^{+0} weeks of gestation.[13] This means that the index condition may have been detected and/or diagnosed before the FA scan. Therefore, the rate for all antenatal detections up to 23^{+0} weeks of gestation is also given.

The 95% confidence intervals (CI) expressing the range of imprecision due to natural variation are calculated using the Wilson score method, which is appropriate for rare events because it yields a range even where the proportion is zero.[14]

2.4 | Data quality control

Data were recorded on NCARDRS' bespoke national database CARA, with front-end input checks to mitigate transcription errors and built-in integrity and completeness rules to flag potential inconsistencies. Every participant pregnancy was audited and interpreted by an NCARDRS registration officer to a standard set of rules. Where information was incomplete, requests were sent to the notifying provider and where this information remained incomplete the participant was excluded from the study. Every participant's information was analysed for data integrity through automated checks of the completeness and validity of both the individual data items and the relationships between them. Every participant's information was also externally validated with the screening provider. Difficult-to-interpret information was queried with expert clinical input, and a random sample of participant pregnancies was moderated by NCARDRS registration officers to ensure standardisation across England. After all data were centrally compiled, providers with outlying low case ascertainment were excluded to mitigate the risk of outcome reporting bias.

2.5 | Patient and public involvement statement

The FASP was introduced by the UK National Screening Committee following consultation with people affected by the conditions screened for and members of the public. This

is a retrospective study of data generated by the programme for the purpose of evaluating the service.

3 | RESULTS

A total of 12 694 participants were identified and audited, shown in Figure 1.

Two groups of participants were no longer eligible by the time the FA scan was due, either because of fetal loss, or termination of pregnancy for another condition ($n = 578$), or because the index condition had already been detected—commonly during the dating scan, by Combined or Quadruple screening for chromosomal conditions, or via private healthcare services ($n = 5811$). Another group of participants either presented to maternity services too late to be scanned, or never booked for antenatal care ($n = 243$). In all, 6009 participants remained eligible for and did not decline the FA scan. The FA scan detected the index condition for 4958 participants, did not detect the index condition for 915 participants, and was inconclusive after two attempts for 80 participants. Fifty-six participants did not receive a complete scan because of gaps in service provision. Condition-specific detection rates and numbers of detections before the FA scan are given in Table 3.

Detection rates of the FA scan met FASP expected detection rates for anencephaly and congenital diaphragmatic hernia, and exceeded them for bilateral renal agenesis, cleft lip ± palate, lethal skeletal dysplasia, spina bifida, and all heart conditions (atrioventricular septal defect, hypoplastic left heart syndrome, tetralogy of Fallot, and transposition of the great arteries). FA scan detection rates did not meet FASP expected detection rates for Edwards syndrome, exomphalos, gastroschisis and Patau syndrome. The majority (78.9%) of detections of these four conditions occurred before the FA scan, and when these are included in the antenatal up to 23^{+0} weeks of detection rate, FASP expected detection rates were met.

4 | DISCUSSION

We report the first national audit of screening performance for the FA scan at 18–23 weeks of gestation. NCARDRS is now able to provide robust national data on the condition-specific detection rates for the scan to inform FASP's national service evaluation and planning, which have formed the basis of recommendations for changes to FASP Screening Standards.[15] All providers were given bespoke reports and linked and enriched record level data on their audited participants for quality assurance and clinical audit. Lower level regional and anonymised provider level intelligence has also been made available for targeted training. This provides evidence of the value of a national register in supporting high-quality clinical care with the population coverage, case ascertainment and expertise to link information that would otherwise exist in isolation.

4.1 | Interpretation

The present study gives test-specific detection rates for the FA scan, within a specific population on a specific

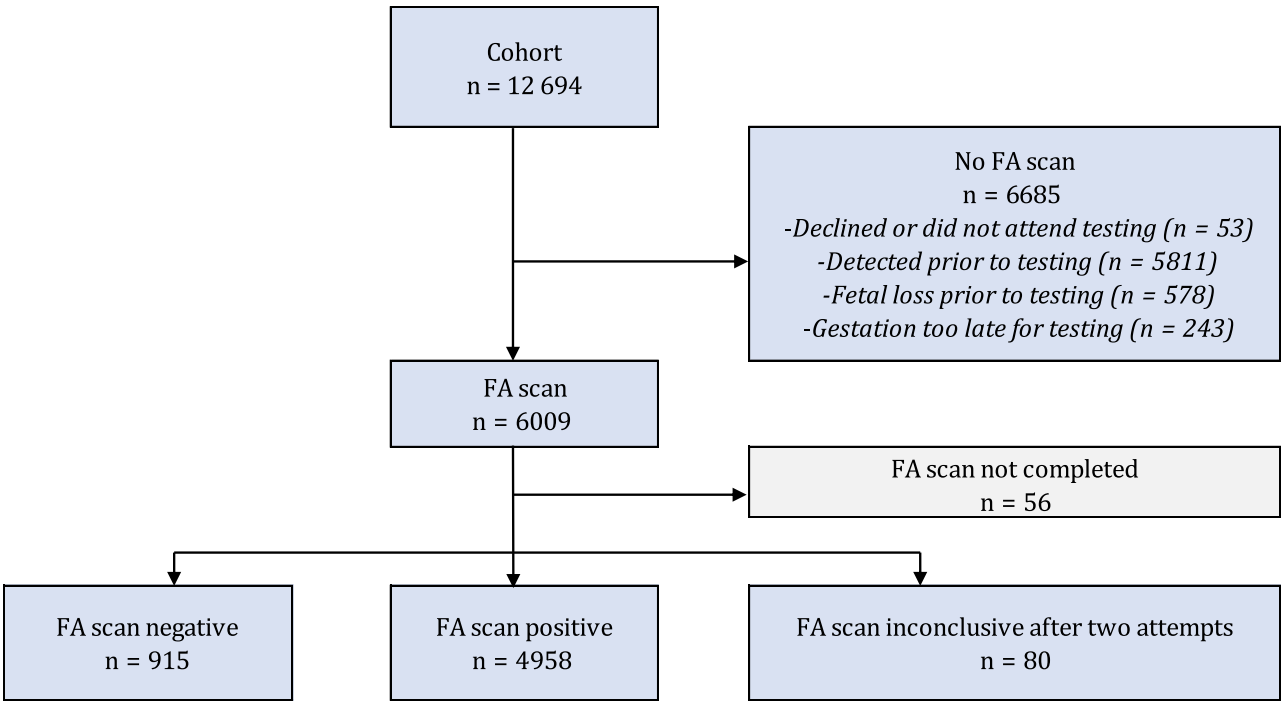

FIGURE 1 Participant flow diagram

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TABLE 3 Fetal anomaly scan detection rates for 14 auditable conditions

Condition	Audited	Detected before the FA scan	Ineligible for or declined FA scan	Detected at FA scan	Undetected, incomplete or inconclusive FA scan	FA scan detection rate (95% CI)	Antenatal up to 23^{+0} weeks detection rate (95% CI)	FASP expected detection rate
Anencephaly	1032	973	32	26	1	96.3% (81.7–99.3%)	99.9% (99.4–100%)	98%
Atrioventricular septal defect	1168	368	59	513	228	69.2% (65.8–72.4%)	79.4% (77–81.7%)	50%
Bilateral renal agenesis	250	66	29	153	2	98.7% (95.4–99.6%)	99.1% (96.8–99.8%)	84%
Cleft lip ± palate	1792	221	118	1300	153	89.5% (87.8–90.9%)	90.9% (89.4–92.1%)	75%
Congenital diaphragmatic hernia	670	128	44	303	195	60.8% (56.5–65%)	68.8% (65.1–72.4%)	60%
Edwards syndrome	1778	1349	219	155	55	73.8% (67.5–79.3%)	96.5% (95.4–97.3%)	95%
Exomphalos	1247	1097	54	57	39	59.4% (49.4–68.7%)	96.7% (95.6–97.6%)	80%
Gastroschisis	581	491	20	62	8	88.6% (79–94.1%)	98.6% (97.2–99.3%)	98%
Hypoplastic left heart syndrome	600	125	21	421	33	92.7% (90–94.8%)	94.3% (92.1–95.9%)	50%
Lethal skeletal dysplasia	278	79	9	177	13	93.2% (88.6–96%)	95.2% (91.9–97.2%)	60%
Patau syndrome	733	488	166	65	14	82.3% (72.4–89.1%)	97.5% (95.9–98.5%)	95%
Spina bifida	1063	288	37	692	46	93.8% (91.8–95.3%)	95.5% (94.1–96.6%)	90%
Tetralogy of Fallot	852	99	43	535	175	75.4% (72.1–78.4%)	78.4% (75.4–81.1%)	50%
Transposition of the great arteries	650	39	23	499	89	84.9% (81.7–87.5%)	85.8% (82.9–88.3%)	50%

screening pathway, plus an alternative detection rate including all antenatal detection up to the end of the 23^{+0} weeks gestational window of the FA scan to account for conditions frequently detected earlier in pregnancy. More commonly, the literature provides rates of overall prenatal detection at any gestation, or within broad gestational windows. Although a comparable albeit smaller population, detection rates given by Boyd in 2011 derived from data collected by the British Isles Network of Congenital Anomaly Registers in 2005 and 2006 are presented as all prenatal diagnoses at any gestation. Conversely, although a study of 11 years of national registrations of transposition of the great arteries in Finland are presented within a specific gestational screening period, the ultrasound protocol is not directly comparable to the FA scan.[16] A systematic review of 11 studies into second-trimester ultrasound detection rates between 1991 and 1998[17] shows test-specific detection rates, but these are hampered by very small numbers and the challenge of active case ascertainment. In the case of a recent systematic review of studies of routine third-trimester ultrasound, the gestational period, although defined, was not directly comparable to the FA scan. Nevertheless it adds valuable information on the proportion of babies who, although undetected at FA scan, may go on to be detected at a later routine scan.[18]

4.2 | Main findings

Compared with FASP expected detection rates (themselves based on a review of 20 years of literature), the FA scan performed as well or better than expected for all conditions other than Edwards syndrome, exomphalos, gastroschisis, and Patau syndrome, which were all significantly below the expected rate. These four conditions, plus anencephaly, were found to be less likely to be eligible for the scan, as the majority of participants experienced early detection or fetal loss before the scan. In April 2015, FASP implemented the offer of screening to all women in England for Edwards syndrome and Patau syndrome, in addition to the existing offer of Down syndrome, at the 11^{+2}–14^{+1} weeks of gestation combined test. The study cohort was the first for whom this offer was uniformly available across England. As a result, the majority (75.9% of Edwards syndrome and 66.6% of Patau syndrome) were detected before the FA scan. Almost all anencephaly and the majority of abdominal wall defects were also detected before the FA scan (anencephaly 94.3%, exomphalos 88.0% and gastroschisis 84.5%). However, the present study did not audit the nature of these early detections and at present there is no standardised protocol for what fetal structures should be examined routinely at the dating scan and therefore detection rates will vary between different

trusts depending on their local protocol. Further study is required to evaluate the potential efficacy of a national first-trimester protocol and NCARDRS are working with Oxford University to provide the data to support a National Institute for Health Research funded health technology assessment study of its potential clinical and cost effectiveness.[19]

4.3 | Strengths and limitations

Auditing the FA scan within the framework of a national screening programme has informed but also restricted the methodology of the audit. First, comparing the accuracy of the index test to established diagnostic reference standards allows for limited critical consideration of the reference standards themselves. As such, where diagnoses were included in the study on the basis of the written report of a consultant or pathologist in the absence of original molecular or imaging data, it was not possible to control for the possibility of inter-individual and inter-institutional variation in diagnosis. Second, aligning the study design with FASP screening standards determined that only singleton and twin pregnancies were eligible for inclusion and higher multiples were excluded.

Third, although the offer of the FA scan is non-selective, the pool of participants eligible at this gestation of pregnancy is the product of selective processes and therefore does not constitute a complete population. Women may decline the FA scan or not book with maternity services in time to be eligible. Pregnancies may be lost before the FA scan, or other conditions may be detected before the FA scan, leading to other care pathways or termination of pregnancy for fetal anomaly. Not only do screening tests and routine scans available at earlier gestations of pregnancy diminish the number of participants still eligible for screening at the FA scan, it is also likely that anatomical features more easily visualised by ultrasound are detected early in pregnancy, leaving those more difficult to detect still eligible for the FA scan, deflating its apparent sensitivity. This may help to explain why the FA scan detection rates for gastroschisis and exomphalos fell below the FASP expected detection rates. As such, no sensitivity calculations were attempted.

This study could not calculate positive predictive value because screening results were only collected for participants with a diagnosis of one of the audited conditions, excluding false positives by design. Likewise, the study could not produce data to calculate either the specificity or the negative predictive value of the FA scan. In the future, NCARDRS needs to be able to provide data on false-positive FA scan results, and a pilot data collection is currently underway to achieve this aim. The study did not collect information on the individuals performing the FA scans or diagnostic testing, or on the scanning or diagnostic technologies used at different providers. Further study is required to understand how inter-institutional variation in implementation of the screening programme affects local detection rates, for example where the literature indicates that consultant-led units achieve higher detection rates.[20]

5 | CONCLUSION

This is the first national audit for the detection of physical conditions at the FA scan and clearly shows the benefit of FASP developing policy, implementing standards and training and NCARDRS providing outcome data. The screening performance is above the expectations set in 2010 for most conditions and the majority of the remainder are detected before the screening window for the FA scan. The unique collaboration between FASP and NCARDRS will inform the modification of expected detection rates, support the study of the potential efficacy of a national first-trimester protocol screening scan for specific conditions and provide the ability to audit local screening performance to support improving detection rates.

AUTHOR CONTRIBUTIONS

NA led the project, conducted the analysis, drafted and revised the paper and is guarantor for the paper. PP clinically led, designed positivity cutoffs and revised the paper. JR academically led and revised the paper. NM led data collection, cleaning and interpretation and revised the paper. JB led data collection, cleaning and interpretation and revised the paper. NP initiated the project, wrote reporting requirements and revised the paper. AM initiated the project, wrote reporting requirements, and revised the paper. SS initiated the project and revised the paper.

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Foundation Trust, Jersey General Hospital Laboratory, Jersey Hospital, Kettering General Hospital Laboratory, Kettering General Hospital NHS Foundation Trust, King George Hospital Laboratory, King's College Hospital Laboratory, King's College Hospital NHS Foundation Trust, Kingston Hospital NHS Foundation Trust, Lancashire Teaching Hospitals NHS Foundation Trust, Leeds Teaching Hospitals NHS Trust, Leicester Royal Infirmary Laboratory, Lewisham and Greenwich NHS Trust, Liverpool Women's NHS Foundation Trust, London North East Thames Regional Genetics Service, London North West Healthcare NHS Trust, London North West Thames Regional Genetics Service, Luton and Dunstable University Hospital NHS Foundation Trust, Maidstone and Tunbridge Wells NHS Trust, Manchester University NHS Foundation Trust (South), Manchester University NHS Foundation Trust (St Mary's), Medway NHS Foundation Trust, Mid Cheshire Hospitals NHS Foundation Trust, Mid Essex Hospital Services NHS Trust, Mid Yorkshire 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Poole Hospital NHS Foundation Trust, Portsmouth Hospitals NHS Trust, Portsmouth Pathology Service, Royal Berkshire NHS Foundation Trust, Royal Cornwall Hospitals NHS Trust, Royal Devon & Exeter Hospital Laboratory, Royal Devon and Exeter NHS Foundation Trust, Royal Free London NHS Foundation Trust, Royal Marsden Cytogenetic Laboratory, Royal Surrey County Hospital NHS Foundation Trust, Royal Sussex County Hospital Laboratory, Royal United Hospitals Bath NHS Foundation Trust, Salisbury NHS Foundation Trust, Sandwell and West Birmingham Hospitals NHS Trust, SE Thames Regional Genetics Laboratories, Severn Pathology, Sheffield Clinical Chemistry Laboratory, Sheffield Teaching Hospitals NHS Foundation Trust, Sherwood Forest Hospitals NHS Foundation Trust, Shrewsbury and Telford Hospital NHS Trust, South Tees Hospitals NHS Foundation Trust, South Tyneside and Sunderland NHS Foundation Trust, South Warwickshire NHS Foundation Trust, Southend University Hospital NHS Foundation Trust, Southport and Ormskirk Hospital NHS Trust, St Georges University Hospital Laboratory, St George's University Hospitals NHS Foundation Trust, St Helens and Knowsley Teaching Hospitals NHS Trust, St James's University Hospital Laboratory, Stockport NHS Foundation Trust, Surrey and Sussex Healthcare NHS Trust, SW Thames Regional Genetics Laboratory, Tameside Hospital NHS Foundation Trust, Taunton and Somerset NHS Foundation Trust, The Doctors Laboratory, The Dudley Group NHS Foundation Trust, The Hillingdon Hospitals NHS Foundation Trust, The Newcastle upon Tyne Hospitals NHS Foundation Trust, The Princess Alexandra Hospital NHS Trust, The Queen Elizabeth Hospital, King's Lynn, NHS Foundation Trust, The Rotherham NHS Foundation Trust, The Royal Bolton Hospital Laboratory, The Royal Bournemouth and Christchurch Hospitals NHS Foundation Trust, The Royal London Hospital Laboratory, The Royal Wolverhampton NHS Trust, The Whittington Hospital NHS Trust, Torbay and South Devon NHS Foundation Trust, UCLH Pathology Laboratory, United Lincolnshire Hospitals NHS Trust, University College London Hospitals NHS Foundation Trust, University Hospital Southampton NHS Foundation Trust, University Hospitals Birmingham NHS Foundation Trust, University Hospitals Bristol NHS Foundation Trust, University Hospitals Coventry and Warwickshire NHS Trust, University Hospitals of Derby and Burton NHS Foundation Trust (Burton), University Hospitals of Derby and Burton NHS Foundation Trust (Derby), University Hospitals of Leicester NHS Trust, University Hospitals of Morecambe Bay NHS Foundation Trust, University Hospitals of North Midlands NHS Trust, Walsall Healthcare NHS Trust, Warrington and Halton Hospitals NHS Foundation Trust, Wessex Regional Genetics Laboratory, West Hertfordshire Hospitals NHS Trust, West Midlands NHS Genomic Medicine Centre, West Suffolk NHS Foundation Trust, Western Sussex Hospitals NHS Foundation Trust, Weston Area Health NHS Trust, Wexham Park Hospital, Wexham Park Hospital Laboratory, Wirral University Teaching Hospital NHS Foundation Trust, Worcestershire Acute Hospitals NHS Trust, Wrightington, Wigan and Leigh NHS Foundation Trust, Wye Valley NHS

Trust, Yeovil District Hospital NHS Foundation Trust, York Hospitals NHS Foundation Trust.

CONFLICT OF INTEREST

None declared. Completed disclosure of interest form available to view online as supporting information.

DATA AVAILABILITY STATEMENT

Study data are not publicly available and access is subject to approval by the UK Health Security Agency Office for Data Release and the NHS Population Screening Programmes' Research Advisory Committee for Antenatal and Newborn Screening.

ETHICS APPROVAL

NCARDRS collected the study data under legal permissions granted under Section 251 of the NHS Act 2006. On 1 October 2021 NCARDRS transferred from Public Health England to NHS Digital and now collects data by Direction of the Secretary of State for Health and Social Care under legal permissions granted under Sections 254(1) and 254(6) of the 2012 Health and Social Care Act. Patients have an absolute right of opt-out from the register and patient information is available online and in print. This work uses data that have been provided by patients, the NHS and other healthcare organisations as part of patient care and support. The data are collated, maintained and quality assured by the National Congenital Anomaly and Rare Disease Registration Service, which is part of NHS Digital.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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