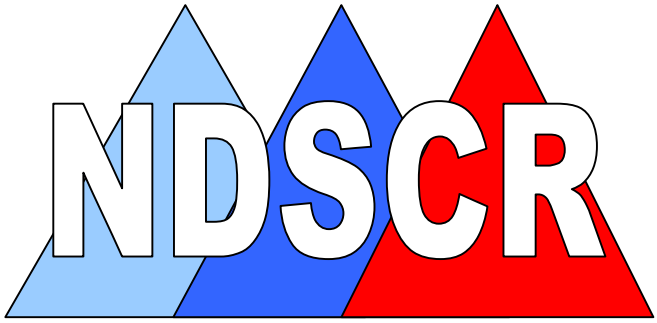
The National Down Syndrome Cytogenetic Register

2002 Annual Report

(funded by the NSC)



Web site: www.smd.qmul.ac.uk/wolfson/ndscr
Phone: 020-7882-6217 / 6220
Fax: 020-7882-6221
Email: ndscr@qmul.ac.uk

Foreword

This 2002 annual report contains information about the NDSCR – who we are and what we do as well as detailed data on all cases of Down syndrome diagnosed cytogenetically from 1989 to 2002.

The NDSCR has undergone several changes over the last year:

- Having previously been funded by NHS Regional Research Funds we are now funded by the National Screening Committee and are working closely with local screening co-ordinators to enable them to carry out their audit function.
- We have expanded to include data on Patau syndrome (trisomy 13) and Edwards syndrome (trisomy 18)
- We have gained PIAG approval through being a member of BINOCAR.
- The role of Director of the register has passed from Eva Alberman to Joan Morris and Wayne Huttly has replaced David Mutton.

We would like to thank all the individuals who contribute to the NDSCR to make it such a valuable resource. We hope that we can continue to count on their collaboration.

Joan Morris – Director NDSCR
Wayne Huttly
Haiyan Wu

December 2003

Contents

Introduction

Welcome to the 2002 annual report of the National Down Syndrome Cytogenetic Register.

The NDSCR is based at The Wolfson Institute of Preventive Medicine, London. It is funded by the National Screening Committee.

Aims of the NDSCR

The NDSCR started in 1989 and aims to collect all cytogenetic or DNA reports of trisomies 21, 13 and 18 and their cytogenetic variants occurring in England and Wales. This data can then be used to help :

- monitor the Down syndrome antenatal screening and diagnostic services and the effect they have on the diagnoses of trisomies 13 (Patau) and 18 (Edwards)
- provide data on annual numbers of affected births to help those planning for their health, educational and social care.
- Provide information for research into the epidemiology of Down, Patau and Edwards syndromes.

How the NDSCR works

All cytogenetic laboratories in England and Wales collaborate with the NDSCR, and provide, on standard forms, a notification of all prenatal and postnatal diagnoses of Down, Patau and Edwards syndromes. (Appendix A gives a list of all 21 laboratories and a copy of the form is shown in Appendix B) The form is self-copying and has 4 pages. The top copy is sent to the NDSCR, the 2nd and 3rd are sent to the referring clinician and the 4th sheet is retained by the laboratory. The clinicians are asked to forward the 3rd copy to the local screening co-ordinator.

The number of Down syndrome pregnancies notified annually has risen from about 1000 in 1989 to over 1400 in 2002, but 2004 is the first year in which notifications of Patau and Edwards syndromes have been requested. We do not yet know how many of these there will be.

What data are collected

The form contains details of the chromosome analysis and some information on the mother and child, including postcode of residence, mother's age, length of pregnancy, the reason for which referral for diagnosis was made, and information about screening. The data do not include full names or addresses, but includes enough information to enable us to identify duplicate registrations.

Data completion and processing

Follow-up of prenatal diagnoses
Only about 8% of prenatal diagnoses of Down syndrome end as a birth. We request the referring physicians to inform us of the pregnancy outcome (birth, termination or miscarriage) and the date and gestational age where a prenatal diagnosis has been made. **No direct contact is ever made with the mothers.**

How the data are stored
The data are entered onto passworded computers in locked offices. The full data are accessible only to the research team.

Validation of data
In order to ensure high levels of completeness, the data are matched with those held by the National Statistics Congenital Anomaly System and some of the Regional Congenital Anomaly Registers. In previous years this has shown the NDSCR data on births to be over 94% complete. Annual lists are sent to the laboratories for them to check that all cases have been registered.

Data quality
The Table in Appendix C gives the proportion of missing data on forms for the years 1989 to 2000 combined and 2001 and 2002 separately. This is always highest in the most recent data where the clinicians have not yet been contacted. Requests for missing data are sent out regularly. The major problem is to ascertain the outcome of prenatally diagnosed pregnancies, particularly where the referral has been from a centre other than that where the mother was booked. Missing data for variables other than outcome are rare, with the exception of the numbers of previous pregnancies, a question that may not be seen as relevant to the clinicians, although it is important in terms of risk of recurrence. There have been many changes in postcodes since the start of the register and the same is true for health authority definitions. Regular recoding is carried out to keep these up-to-date.

Speed of reporting
Although most laboratories provide data within six months of the diagnoses we are hopeful that the involvement of the National Screening Committee and local screening co-ordinators will speed up the provision of outcome data, and the provision of more complete information on pregnancy history.

Data confidentiality and informed consent

Personal information held on a computer system is safeguarded by the Data Protection Act 1998 and the NDSCR is registered under this.

The Government has made it clear that informed consent is a fundamental principle governing the use of patient identifiable information. However it also recognises that situations may arise where informed consent cannot practically be obtained. Section 60 of the Health and Social Care Act 2001 provides a power to ensure that patient identifiable information needed to support essential NHS activity can be used without the consent of patients. The Act requires that the National Patient Advisory Group (PIAG) consider applications to use patient identifiable information without full informed consent. During 2003 the NDSCR has worked with the British Isles Network of Congenital Anomaly Registers (BINOCAR) to submit an application to PIAG to allow the register to operate without informed consent whilst consent procedures are developed wherever possible. This application was successful.

How the data are used

Audit of Down Syndrome Screening

- All local screening co-ordinators should receive the green section of the NDSCR form which will asssit them in their audit requirements to.
- Annual reports are produced describing numbers of prenatal and postnatal diagnoses, and the methods of prenatal screening which led to prenatal diagnoses.
- More detailed information is regularly published in medical journals.

Feedback

- NDSCR leaflets giving information on the trends in DS diagnosis are produced annually and distributed to cytogenetic laboratories, local screening co-ordinators and clinicians.
- The NDSCR web site is regularly updated.

Recent special studies

In-house studies

1) To test the general validity of our previous findings showing that the age related risk of a DS pregnancy is not as great in women over 45 as was previously assumed (Morris et al 2002), we are analysing the corresponding data from registries who are members of EUROCAT. (European Concerted Action on Congenital Anomalies and Twins)
2) We are investigating the peak of Down syndrome cases occurring in 1997. This peak cannot be explained by the increase in pregnancies in 1997, the maternal age distribution or the number of prenatal diagnoses.
3) We are using data on mothers who have a history of more than one Down syndrome pregnancy to develop newer and simpler estimates of maternal age-specific recurrence risk.

Collaborative studies

1) David Neasham from SASHU (Small Area Health Statistics Unit) has used relevant NDSCR data for his study which shows residence in wards designated as low social class appears to increase the risk of a Down syndrome pregnancy in mothers under 35. The opposite social class gradient exists in older mothers (Neasham, D. PhD; London University 2003).

2) We are continuing our collaboration with the National Childhood Cancer Register, to estimate the age-specific risk of leukaemia in children with DS, where we are able to provide denominator data for children on their register.

3) We are collaborating with Dr Kovaleva from St Petersburg who is studying the epidemiology of double aneuploidy involving trisomy 21.

4) We have helped Dr Jill Ellis of the Institute of Child Health with a study of the effect of special diets on the development of children with Down syndrome.

A list of publications based on, or using NDSCR data are given in Appendix D.

New data to be collected: Patau and Edwards syndromes

The increase in prenatal screening for Down syndrome has resulted in an increase in the numbers of prenatally diagnosed cases of Patau and Edwards syndromes. The National Screening Committee has agreed that the NDSCR should start collecting data on these diagnoses. We have specified that this extra information is to be recorded on the same form as the Down syndrome notifications and have revised the form accordingly.

The Data in the NDSCR

Down syndrome cases diagnosed in 2002

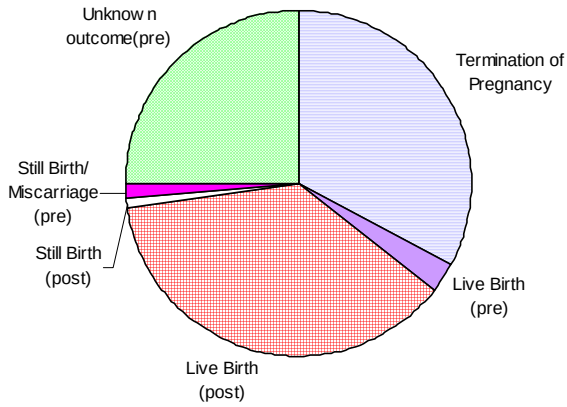
There were 1433 Down syndrome diagnoses made in 2002, 886 (62%) made prenatally and 547 (38%) postnatally (Table 1 and Figure 1). If we assume that of the prenatal diagnoses with unknown outcomes the proportion terminated remains unchanged, the likely number of Down syndrome live births in England and Wales in 2002 would have been 599 (37 + 533 + 8% of 359), a birth prevalence of 1 per 1000 livebirths.

Table 1: Down syndrome cases diagnosed in 2002 by time of diagnoses and outcome

Time of Diagnoses	Outcome	Number	(%)
Prenatal	Termination of pregnancy	471	(33)
	Live Birth	37	(3)
	Still Birth / Miscarriage	19	(1)
	Unknown outcome†	359	(25)
Postnatal	Live Birth	533	(37)
	Still Birth	14	(1)
Total		1433	(100)

†: About 8% of those with unknown outcomes are likely to result in a live birth.

Figure 1: Down syndrome diagnoses in 2002 (pre=prenatal diagnosis, post=postnatal diagnosis)



Indication for prenatal karyotyping

The indications for karyotyping reflect the different methods of prenatal screening occurring. In 58% of all prenatally diagnosed cases the indication mentioned was the result of an ultrasound scan. The majority of these were carried out before 20 weeks, and were probably nuchal translucency measurements. Unfortunately we cannot determine this precisely from the form used in 2002. This information should be available for 2004 from the revised form. In 29% of prenatally diagnosed cases the indication was a maternal serum test, in 16% it was maternal age and in 1% a previous Down syndrome pregnancy. More than one reason can be given and therefore the reasons do not add up to 100%.

Gestational age at prenatal diagnoses

Of the 886 prenatally diagnosed cases, 21% were diagnosed before 13 weeks, 67% before 17 weeks and only 8% over 20 weeks gestation (Table 2). This pattern reflects the type of screening that had led to the prenatal diagnosis.

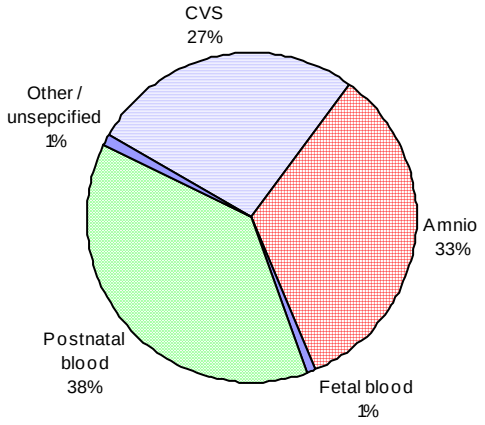
Table 2: Down syndrome cases diagnosed prenatally by gestational age at diagnoses in 2002

Gestational age weeks)	DS cases No. (%)
< 13	192 (21)
13 - 14	204 (23)
15 - 16	202 (23)
17 - 18	157 (18)
19 - 20	62 (7)
21 +	69 (8)
Total	886 (100)

Tissue used for karyotyping

The most common source of method of sampling fetal cells in 2002 was amniocentesis, with chorionic villus sampling being almost as common (Figure 2). The median time from CVS sampling to termination of pregnancy was 7 days compared with 11 days for amniocentesis. 87% of all terminations following CVS were within 14 days of the procedure compared with 60% for amniocentesis.

Figure 2: Tissue used for karyotyping in 2002



Maternal age at diagnoses

The average age of the mother at the time of diagnosis of fetal Down syndrome was 35; the most common age was 39 years (Figure 3) and a third were between 35 and 39 years old (Table 3).

Regional differences in Down syndrome cases diagnosed in 2002

Tables 4 and 5 show the patterns of diagnoses of Down syndrome across England and Wales. The proportion of cases diagnosed prenatally varies from 45% in Northern & Yorkshire to 76% in Eastern NHS region. The completeness of the data also varies by region with only 7% of outcomes being unknown in West Midlands compared to 42% in Wales. Table 5 shows that women in the regions with a higher proportion of referrals due to an ultrasound scan (probably mostly nuchal translucency measures in the first trimester) are more likely to have had a CVS than an amniocentesis.

Figure 3: Maternal age at diagnoses in 2002

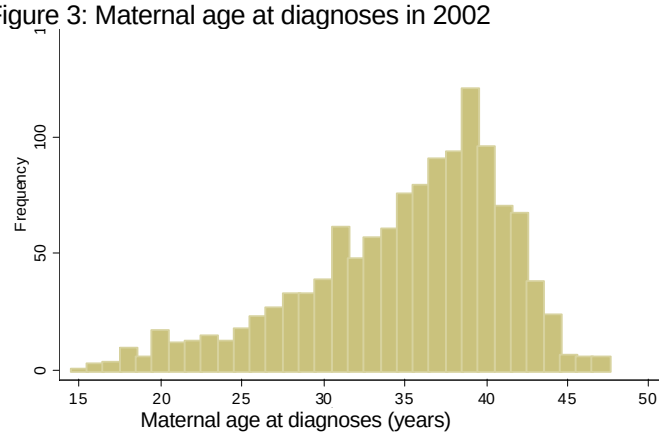


Table 3: Maternal age at diagnoses in 2002

Maternal age (years)	DS cases No. (%)
< 20	24 (2)
20 -	70 (5)
25 -	134 (9)
30 -	267 (19)
35 -	462 (32)
40 -	297 (21)
45 +	19 (1)
missing age	160 (11)
Total	1433 (100)

Table 4: Down syndrome diagnoses by NHS Regional Office Area in 2002

Regional Office Area	Number of Down Syndrome diagnoses	Prenatal diagnoses (%)	Median maternal age at diagnosis (years)	Missing [+] maternal age (%)	Unknown [+] outcomes (%)
North & Yorkshire	165	45	36.5	15	12
Trent	96	56	34.7	5	8
West Midlands	129	53	35.0	16	7
North West	153	58	37.6	12	21
Eastern	129	76	36.0	9	36
London	327	66	37.5	15	39
South East	244	70	36.9	7	31
South West	130	58	37.0	6	10
Wales	60	63	35.6	10	42
Total	1,433	62	36.8	11	25

[+]: Data currently being updated.

Table 5: Prenatal diagnoses by NHS Regional Office Area, indication and tissue used for diagnosis in 2002

Regional Office Area	Number of diagnoses	Indications for karyotyping mentioned*			Median Gestational age at sample (weeks)	Proportion of tissue from	
		Ultrasound scan (%)	Serum test (%)	Age (%)		CVS	Amnio-centesis
North & Yorkshire	75	65	17	27	15	59	36
Trent	54	37	41	15	16	33	63
West Midlands	68	28	50	27	17	18	81
North West	89	43	34	34	16	11	89
Eastern	98	53	41	7	16	35	61
London	216	65	26	9	13	64	34
South East	172	73	19	11	13	50	49
South West	76	61	28	11	15	49	46
Wales	38	53	34	32	16	11	84
Total	886	58	29	16	15	43	54

* More than one indication may be mentioned

Trends over time in Down syndrome Diagnoses

Since the register started collecting data on 1st January 1989 there has been a dramatic increase in the proportion of Down syndrome cases detected prenatally from 30% in 1989 to 62% in 2002, and numbers detected prenatally from 321 to 886 in 2002. (Table 6 and Figure 4). There is a corresponding decrease in the numbers of live births reported although this trend is now plateauing.

The total number of Down syndrome diagnoses has increased steadily due to the trend of women to have their children later in life and also the increase in earlier prenatal diagnoses and subsequent terminations. Many of these fetuses diagnosed prenatally would have miscarried naturally. Therefore although the numbers of Down syndrome diagnoses are rising annually, the maternal age-related risk of having a Down syndrome birth has remained constant since 1989.

There was an increase in the proportion of mentions of a serum test as an indication for karyotyping from only 6% in 1989 to just under 40% from 1992 to 1997 (Table 7). This proportion then decreased with the introduction of nuchal translucency measurements as a screening test. In 2002 a serum test was mentioned as an indication for prenatal diagnosis in 29%, 58% mentioning ultrasound results as an indication. The use of maternal age as an indication for karyotyping is decreasing steadily, although even in 2002 it was given as an indication in 16% of prenatal diagnoses.

As the screening tests are being done at earlier gestations, an increasing number of women are having chorionic villus sampling (CVS) instead of amniocentesis, the ratios being 18% CVS to 77% amniocentesis in 1989, and 43% to 54% respectively in 2002. (Table 7)

Figure 4: The number of prenatal and postnatal diagnoses by year of diagnosis

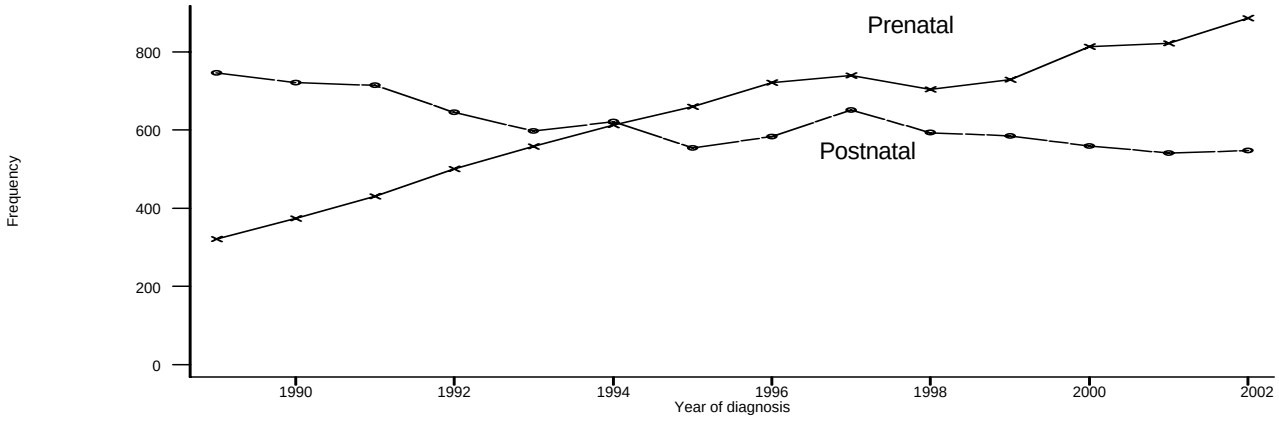


Table 6: Down syndrome diagnoses and outcomes in England and Wales from 1989 to 2002

Year of Diagnoses	Number of Down syndrome diagnoses	Proportion of prenatal diagnoses %	Number of Live Births	Number of Terminations	Number of Miscarriages* and Still Births	Number of Unknown Outcomes
1989	1,067	30	750	293	16	8
1990	1,095	34	738	327	17	13
1991	1,144	38	735	369	31	9
1992	1,145	44	661	442	24	18
1993	1,155	48	622	506	18	9
1994	1,234	50	638	541	29	26
1995	1,214	54	579	578	32	25
1996	1,304	55	606	651	31	16
1997	1,390	53	667	654	40	29
1998	1,297	54	629	607	21	40
1999	1,314	55	603	635	31	45
2000	1,372	59	590	621	24	137
2001	1,363	60	570	528	20	245
2002	1,433	62	570	471	33	359
Total	17,527	51	8,958	7,223	367	979

* Only miscarriages after prenatal diagnosis are included

Table 7: Down syndrome prenatal diagnoses 1989 to 2002

		Indications for Karyotyping mentioned*				Proportion of tissue from	
Year of diagnoses	Number of diagnoses	Ultrasound scan %	Serum test %	Mat. Age %	Gestational age at sample (weeks)	CVS %	Amnio-centesis %
1989	321	13	6	79	16	18	77
1990	374	17	15	71	16	16	76
1991	430	22	22	59	17	15	73
1992	500	27	37	47	17	11	79
1993	558	28	40	42	17	17	77
1994	613	30	41	37	17	23	69
1995	660	40	37	34	16	25	69
1996	721	40	38	30	16	30	65
1997	739	48	38	22	16	35	61
1998	704	51	31	24	16	35	61
1999	729	51	34	20	16	33	60
2000	813	56	33	18	16	37	59
2001	822	61	26	20	15	45	52
2002	886	58	29	16	15	43	54
Total	8,870	43	32	32	16	30	64

* More than one indication may be mentioned

Appendix A

List of Cytogenetic Laboratories in England and Wales

NORTHERN GENETICS SERVICE
Institute of Human Genetics, International Centre For Life, Central Parkway, Newcastle Upon Tyne. NE1 3BZ Tel: 0191 241 8700

REGIONAL GENETIC SERVICE, MANCHESTER
St Mary's Hospital, Hathersage Road, Manchester, M13 OJH Tel: 0161 276 6553

ROYAL MANCHESTER CHILDREN'S HOSPITAL
Hospital Road, Pendlebury, Manchester, M27 4HA Tel: 0161 727 2567

CHESHIRE AND MERSEYSIDE GENETICS SERVICE
Liverpool Women's Hospital, Crown Street, Liverpool, L8 7SS Tel: 0151 702 4229

YORKSHIRE REGIONAL GENETICS SERVICE
Ashley Wing, St James's University Hospital, Beckett Street, Leeds, LS9 7TF
Tel: 0113 206 5550

NORTH TRENT GENETIC SERVICES
Sheffield Children's Hospital, Western Bank, Sheffield, S10 2TH Tel: 0114 271 7015

NOTTINGHAM GENETIC SERVICE
City Hospital, Hucknall Road, Nottingham, NG5 1PB Tel: 0115 962 7617

LEICESTERSHIRE GENETICS CENTRE
University Hospitals of Leicester NHS Trust, Leicester Royal Infirmary, Leicester, LE1 5WW Tel: 0116 258 5637

WEST MIDLANDS REGIONAL GENETICS SERVICE
Birmingham Women's Hospital, Edgbaston, Birmingham, B15 2TG Tel: 0121 627 2710

OXFORD REGIONAL GENETICS SERVICE
Oxford Radcliffe Hospitals NHS Trust, The Churchill, Old Road, Headington, Oxford, OX3 7LJ Tel: 01865 226 001

EAST ANGLIAN REGIONAL GENETICS SERVICE
Regional Genetics Laboratories, Kefford House, Maris Lane, Trumpington, Cambridge, CB2 2FF Tel: 01223 550 700

NORWICH MOLECULAR AND CYTOGENETICS SERVICE
Norfolk and Norwich University Hospital, Colney Lane, Norwich NR4 7UY Tel: 01603 286 038

SOUTH WESTERN REGIONAL GENETICS SERVICE
St Michael's Hospital, Southwell Street, Bristol BS2 8EG Tel: 0117 959 5570

NW THAMES REGIONAL GENETICS SERVICE
(Kennedy-Galton Centre) NWLH NHS Trust Level 8v, Watford Road, Harrow, Middlesex, HA1 3UJ Tel: 020 8869 3154

NE THAMES REGIONAL GENETICS SERVICE
Clinical Genetics Unit, Institute of Child Health, 30 Guilford Road, London, WC1N 1EH
Tel: 020 7829 8870

SW THAMES REGIONAL GENETICS CENTRE
Medical Genetics Unit, St George's Hospital Medical School, Cranmer Terrace, London, SW17 0RE Tel: 020 8725 5332

GUY'S AND ST THOMAS' HOSPITAL NHS TRUST
Cytogenetics Department, 5th Floor, Guy's Tower, Guy's Hospital, London, SE1 9RT
Tel: 020 7955 8719

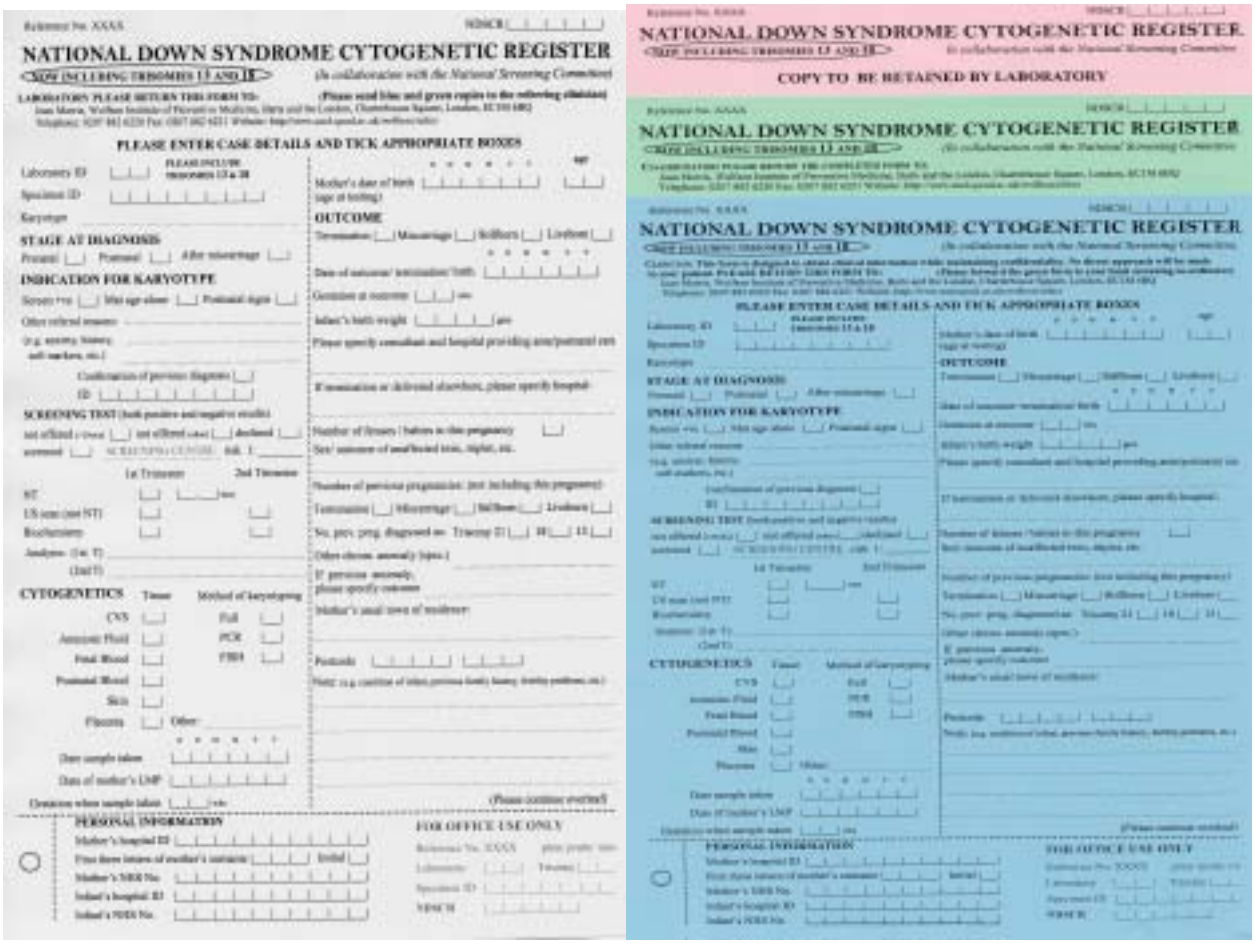
WESSEX CLINICAL GENETICS AND LABORATORY SERVICE
Salisbury District Hospital, Salisbury, Wiltshire, SP2 8BJ Tel: 01722 429080

WALES
Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff, CF14 4XW
Tel: 02920 744 054

CYTOGENETICS SERVICES
(TDL Genetics as from 20/02/04)
3rd Floor North, 60 Whitfield Street, London W1T 4EU Tel: 020 7486 1322

Appendix B

New form


NATIONAL DOWN SYNDROME CYTOGENETIC REGISTER

NATIONAL DOWN SYNDROME CYTOGENETIC REGISTER

COPY TO BE RETAINED BY LABORATORY

NATIONAL DOWN SYNDROME CYTOGENETIC REGISTER

Appendix C

Data Completeness

The following table gives the completeness of different data items for the years 1989 to 2000 and 2001 and 2002. We are still following up the missing data for 2001 and 2002. The data from 1989 to 2000 is included for comparison purposes to demonstrate the levels of completeness we are aiming to achieve for the 2001 and 2002 data.

Table C1: Completeness of data from 1989 to 2002

	Percentage complete		
	1989-2000	2001	2002
Data Item			
Reason for referral for karyotyping	100%	100%	99%
Type of tissue karyotyped	100%	99%	100%
Sex of fetus	100%	100%	98%
Maternal age	97%	90%	89%
Gestational age at sample for prenatal diagnoses	100%	100%	100%
Outcome of pregnancy	97%	82%	75%
Gestational age at outcome for prenatal diagnoses	84%	64%	55%
Number of previous pregnancies	66%	58%	52%
Post Codes (some information)	92%	94%	95%
(complete postcodes)	84%	85%	84%

Appendix D

NDSCR Publications

1. Mutton DE, Alberman E, Ide R, Bobrow M. Results of first year (1989) of a national register of Down's syndrome in England and Wales. *BMJ* 1991; **303**:1295-7
2. Mutton DE, Ide R, Alberman E, Bobrow M. Analysis of National Register of Down's syndrome in England and Wales: trends in prenatal diagnosis. *BMJ* 1993; **306**: 431-2.
3. Morris JK, Mutton DE, Ide R, Alberman E, Bobrow M. Monitoring trends in prenatal diagnosis of Down's syndrome in England and Wales, 1989-1992. *J Med Screening* 1994; **1:** 233 -7.
4. Alberman E, Mutton D, Ide R, Nicholson A, Bobrow M. Down's syndrome births and pregnancy terminations in 1989 to 1993: preliminary findings. *Br J Obst Gyn*.1995; **102**; 445-7.
5. Huang T, Watts HC, Wald NJ, Morris JK, Mutton D, Alberman E. Reliability of statistics on DS notifications. *J Med Screen* 1997; **4**: 94-97. Subsequent letter in same journal.
6. Williamson P, Harris R, Church S, Fiddler M, Rhind J.Prenatal genetic services for Down's syndrome: access and provision. Steering Committee of the National Confidential Enquiry into Counselling for Genetic Disorders. *Br J Obstet Gynaec* 1996; **103:**676-83.
7. Smith-Bindman R, Waters J, Mutton D, Alberman E. Trends in the effectiveness and efficiency of prenatal Down syndrome (DS) screening in England and Wales, 1989-1999. J Med Genet 2001: Supplement 1 SP33.
8. Mutton DE, Alberman ED, Hook EB. (The National Down Syndrome Cytogenetic Register and the Association of Clinical Cytogeneticists) Cytogenetic and epidemiological findings in Down syndrome: 1993. *J Med Genet* 1996; **33**: 387-394.
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10. Morris JK, Wald NJ, Watt HC. Fetal loss in Down's syndrome pregnancies. Prenatal Diagnosis.1999; **19**:142-145
11. Morris JK, Alberman E, Mutton D. Is there evidence of clustering in Down syndrome? *Int J Epidem* 1998; **27**: 495-498.
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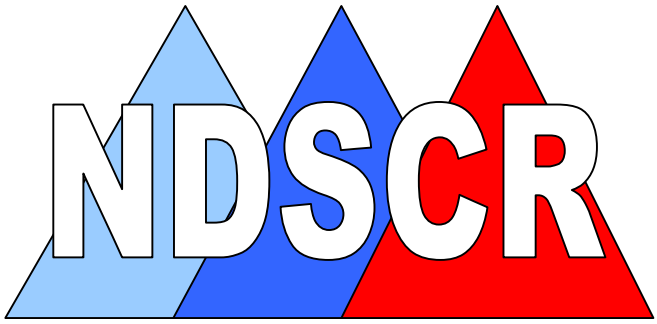
13. Hook EB, Cross PK, Mutton DE. Female predominance (low sex ratio) in 47, +21 mosaics. Am J Med Genet. 1999; **84**:316-319.
14. Vrijheid M, Dolk H, Armstrong B, Abramsky L, Bianchi F, Fazarinc I, Garne E, Ide R, Nelen V, Robert E, Scott JES, Stone D, Tenconi R. Chromosomal congenital anomalies and residence near hazardous waste landfill sites. Lancet 2002, **359**, 320-3.
15. Morris JK, Mutton DE, Alberman E. Revised estimates of the maternal age specific live birth prevalence of Down's syndrome. J Medical Screening, 2002, **9**,2-6
16. Alberman E. The National Down Syndrome Cytogenetic Register (NDSCR) J Medical Screening 2002, **9**, 97-98 (Editorial)
17. Morris JK, Wald NJ, Mutton DE, Alberman E. Comparison of models of maternal age-specific risk for Down syndrome live births. Prenat Diagn. 2003,**23**:252-8.
18. Smith-Bindman R, Chu P, Bacchetti P, Waters JJ, Mutton D, Alberman E. Prenatal screening for Down syndrome in England and Wales and population-based birth outcomes. Am J Obstet Gynecol. 2003; 189(4): 980-5.
19. Alberman E, Huttly W, Hennessy E, McIntosh A. The use of record linkage for auditing the uptake and outcome of prenatal serum screening and prenatal diagnostic tests for Down syndrome. Prenat Diagn. 2003 Oct; 23(10): 801-6.

Conclusions

- The NDSCR has continued to maintain a near complete record of all Down syndrome diagnoses in England and Wales in 2002
- The NDSCR has gained support under Section 60 of the Health and Social Care Act 2001.
- In 2002 there were 1,433 diagnoses of Down syndrome, of which 62% were prenatally diagnosed.
- In 2002 the Down syndrome live birth rate was around 1 per 1000 (this figure is provisional as there are a large number of missing outcome forms).
- At present the large number of missing outcome forms is unacceptable. We hope that by working with the local screening co-ordinators we will be able to reduce this and prevent it from occurring in future years.
- The NDSCR is now funded by the National Screening Committee and has expanded to collect Patau and Edwards syndrome diagnoses in addition to Down syndrome. It is working with the regional and local screening co-ordinators to help them fulfil their audit function.



(funded by the NSC)

Wolfson Institute of Preventive Medicine
Barts and The London
Queen Mary's School of Medicine and Dentistry
Charterhouse Square
London, EC1M 6BQ

Web site: www.smd.qmul.ac.uk/wolfson/ndscr
Phone: 020-7882-6217/6220
Fax: 020-7882-6221
Email: ndscr@qmul.ac.uk