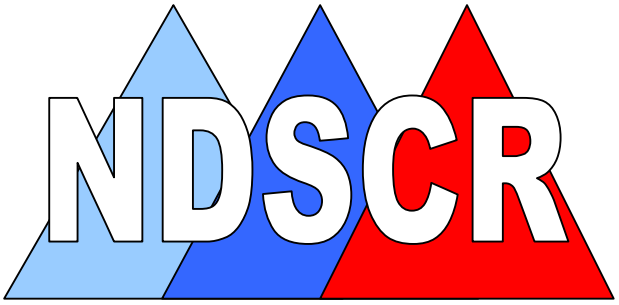
The National Down Syndrome Cytogenetic Register

2004 Annual Report

(data collection funded by the National Screening Committee)



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Foreword

This 2004 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 2004.

The NDSCR has undergone several changes over the last year:

- This is the first annual report that will include a full year's data on diagnoses of Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13) which we began collecting in November 2003.

- We have re-applied for and been successful in obtaining ethical approval to continue collecting our data (from the Trent MREC and PIAG) through being a member of BINOCAR.

- NDSCR Steering Committee
  The following members of the NDSCR steering committee met for the first time in May 2005:
  Dr Joan Morris (Chair – Director NDSCR)
  Dr Jenny Kurinczuk
  Professor Charles Rodeck
  Ms Susannah Seyman
  Dr Jonathan Waters

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
Eva Alberman
David Mutton
Annabelle Stapleton
Haiyan Wu

December 2005

Contents

Introduction

Welcome to the 2004 annual report of the National Down Syndrome Cytogenetic Register.

The NDSCR is based at the Centre for Environmental and Preventive Medicine, Wolfson Institute of Preventive Medicine, Queen Mary's in London. The register is funded by the National Screening Committee.

Aims of the NDSCR

The NDSCR was started in 1989 and we aim to collect all cytogenetic or DNA reports of trisomies 21, 18 and 13 and their cytogenetic variants occurring in England and Wales. These 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 18 (Edwards syndrome) and 13 (Patau syndrome);
- 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, Edwards and Patau 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, Edwards and Patau 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 by the laboratory, the 2nd (blue) and 3rd (green) are sent to the referring clinician and the 4th (pink) sheet is retained by the laboratory. The clinicians are asked to forward the 3rd (green) copy to the local screening co-ordinator, who is usually based within the Antenatal Unit at referring hospital.

The number of Down syndrome pregnancies notified annually has risen from around 1000 in 1989 to 1659 cases in 2004. In November 2003 we first requested notifications of Edwards and Patau syndromes. A total of 151 diagnoses of Patau syndrome and 341 diagnoses of Edwards syndrome were recorded in 2004.

What data are collected

The notification 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 referral for diagnosis and antenatal screening information. To preserve anonymity, the data do not include full names or addresses, but include enough information to enable us to identify duplicate registrations.

Data completion and processing

Follow-up of prenatal diagnoses
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 by the NDSCR.**

The data we have on outcome show that after the prenatal diagnosis of Down syndrome 94% of affected pregnancies are legally terminated and 6% are continued, some miscarrying naturally and some ending as stillbirths. There is often a time lapse before we are informed of these outcomes (see below).

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

Validation of data
In order to ensure high levels of ascertainment, 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 2001 combined; and separately for 2002, 2003 and 2004. 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 provide 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 Act.

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 arise where informed consent cannot practicably 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 Information Advisory Group (PIAG) consider applications to use patient identifiable information without full informed consent. In 2003, and again in 2004, the NDSCR as a part of the British Isles Network of Congenital Anomaly Registers (BINOCAR) was given permission to operate without informed consent. In 2004 the application of the NDSCR for ethics approval from the Trent multi-centre research ethics committee (MREC), as part of BINOCAR, was also approved.

How the data are used

Audit of Down Syndrome Screening

- All local screening co-ordinators should receive the green copy of the NDSCR form to assist them in their audit requirements.
- 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 Down syndrome 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) Using data from the register on mothers who have a history of more than one Down syndrome pregnancy we have estimated the recurrence risk of a mother according to the age at which she had her first Down syndrome pregnancy. (Morris et al, 2005)

2) By combining data from the NDSCR and data from registries who are members of EUROCAT (European Concerted Action on Congenital Anomalies and Twins) we are investigating the risk of a woman having a Down syndrome pregnancy given that she has already had a pregnancy affected with trisomy 13 or 18.

3) We have demonstrated that the risk of natural fetal loss in Down syndrome pregnancies increases with the age of the mother more steeply than this risk in chromosomally normal pregnanies.

Collaborative studies

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

2) We have helped Dr Jill Ellis of the Institute of Child Health, Great Ormond Street Hospital for Sick Children, with a study of the effect of special diets on the development of children with Down syndrome.

Future studies

1) In 2004 we started collecting data on whether women had been offered screening and had accepted or rejected the offer. Once more data is available we will be reporting on the efficacy of Down syndrome in England and Wales.

2) In 2004 there is a large increase in prenatal diagnoses compared to 2003, reasons for this increase are being investigated.

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

The Data in the NDSCR

Down syndrome cases diagnosed in 2004

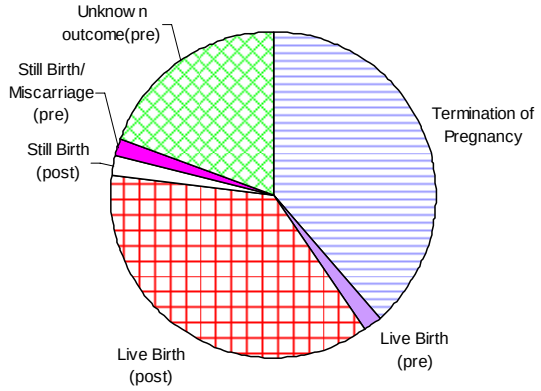
There were 1659 Down syndrome diagnoses made in 2004, 1021 (62%) prenatally and 638 (38%) postnatally (Table 1 and Figure 1). There were 323 prenatal diagnoses of which the outcome is as yet unknown. Assuming that their proportion terminated remains as before 2004, the likely number of Down syndrome live births in England and Wales in 2004 would have been 657 (32+ 606 + 6% of 323), a prevalence of 1 per 1000 in the livebirths occurring in England and Wales in 2004.

Table 1: Down syndrome cases diagnosed in 2004* by time of diagnoses and outcome

		No.	%
Prenatal	Termination of pregnancy	640	39
	Live Birth	32	2
	Still Birth / Miscarriage	26	2
	Unknown outcome†	323	19
Postnatal	Live Birth	606	37
	Still Birth	32	2
Total		1659	100

* 2004 data are provisional.
† About 6% of those with unknown outcomes are likely to result in a live birth.

Figure 1: Down syndrome diagnoses in 2004*
(pre= prenatal diagnosis, post = postnatal diagnosis)



* 2004 data are provisional.

Indication for prenatal karyotyping

The indications for karyotyping reflect the occurrence of different methods of prenatal screening (Figure 3). In 39% of all prenatally diagnosed cases the indication mentioned was the result of an early ultrasound (This is likely to have been an nuchal translucency (NT) measurement), in 18% it was a serum screening test result and in 5% it was both

Gestational age at prenatal diagnoses

Of the 1021 prenatally diagnosed cases, 25% were diagnosed before 13 weeks, 70% before 17 weeks and only 7% 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 according to gestational age at diagnoses in 2004*

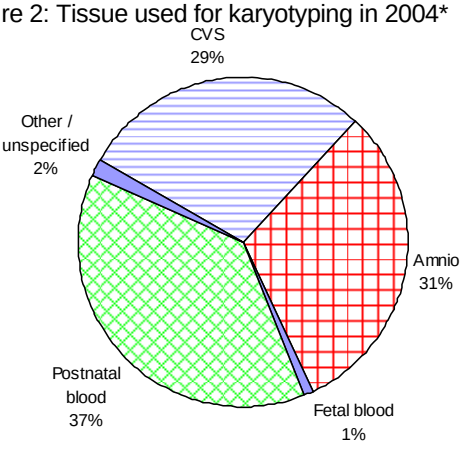
Gestational age (wks)	No.	%
<13	256	25
13-	221	22
15-	239	23
17-	162	16
19-	67	7
21+	76	7
Total	1021	100

* 2004 data are provisional.

Tissue used for karyotyping

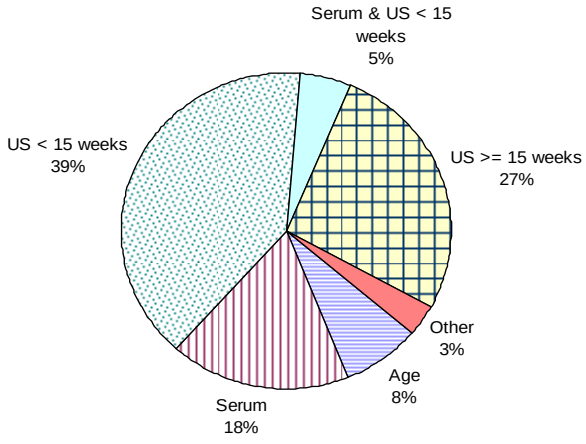
Although in 2004 amniocentesis remained the most common method of sampling fetal cells, chorionic villus sampling was almost as common (Figure 2). The median time from CVS sampling to termination of pregnancy was 7 days compared with 9 days for amniocentesis. 89% of all terminations following CVS were within 14 days of the procedure compared with 77% for amniocentesis.

Figure 2: Tissue used for karyotyping in 2004*



*****2004 data are provisional.

Figure 3: Indication for prenatal karyotyping in 2004*



*2004 data are provisional.

Maternal age at diagnosis

The mean age of the mother at the time of diagnosis of fetal Down syndrome was 36 and 39% (566/1451) of the mothers of known age were between 35 and 39 years (Table 3).

Table 3: Down syndrome cases diagnosed in 2004* according to maternal age at diagnosis

Maternal age (years)	No.	%
<20	28	2
20-	70	4
25-	109	7
30-	330	20
35-	566	34
40-	323	19
45+	25	2
missing	208	12
Total	1659	100

* 2004 data are provisional.

Patau and Edwards syndrome cases diagnosed in 2004

As expected over 90% of both the Patau and Edwards syndrome diagnoses were made prenatally (Table 4), with only a small proportion of all diagnoses being live births.

Table 4: Patau and Edwards syndrome cases by time of diagnoses and outcome in 2004*.

	Syndrome			
	Patau	No.(%)	Edwards	No. (%)
Termination (pre)	100	(66)	184	(54)
Live Birth(pre)	1	(1)	3	(1)
Still Birth/ Mis(pre)	6	(4)	13	(4)
NK[+] outcome(pre)	33	(22)	111	(33)
Live Birth(post)	10	(7)	29	(9)
Still Birth(post)	1	(1)	1	(0)
Total	151	(100)	341	(100)

* 2004 data are provisional.
[+] NK: unknown.

The main indication for karyotyping was an ultrasound scan (excl NT) (around one half) or an NT measurement (around one third) (Table 5).

Table 5: Prenatally diagnosed Patau and Edwards syndrome cases in 2004: Percent of different indications for karyotyping

Indication for Karyotyping	Syndrome	
	Patau (%)	Edwards (%)
NT measurement (may include serum)	30	36
Serum screening (excl. NT)	6	8
Ultrasound (excl. NT)	53	51
Maternal age alone	6	4
Other	5	1
Total	100	100

* 2004 data are provisional.

Regional differences in cases diagnosed in 2004

Table 6 shows the patterns of diagnoses of Down syndrome across England and Wales. The proportion of cases diagnosed prenatally varies from 37% in North East GRO to 69% in London. Women in the regions with a higher proportion of referrals due to an ultrasound scan before 15 weeks (probably nuchal translucency measures in the first trimester) were more likely to have had a CVS than an amniocentesis.

Trends over time in Down syndrome Diagnoses

Since the register started collecting data on 1st January 1989 the total number of Down syndrome diagnoses has increased steadily partly due to increasing maternal age and partly because of the increase in prenatal diagnosis. The proportion diagnosed prenatally has risen from 30% in 1989 to 62% in 2004, and the numbers from 321 to 1021 in 2004. (Table 7 and Figure 4) Since the rate of natural fetal loss in Down syndrome is very high, the potential losses in those diagnosed and subsequently terminated early must be adjusted for before looking at the maternal age-related risk and having a Down syndrome birth. When this is done it is evident that 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 1993 to 1996 (Table 8). This proportion then decreased with the introduction of nuchal translucency measurements as a screening test. In 2004 a serum test was mentioned as an indication for prenatal diagnosis in 18%, 40% mentioning an ultrasound before 15 weeks and 5% mentioning both. NT measurements (with or without serum screening) as an indication. The use of maternal age alone as an indication for karyotyping is decreasing steadily, and in 2004 it was given as an indication in only 8% 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 47% to 51% respectively in 2004. (Table 8)

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

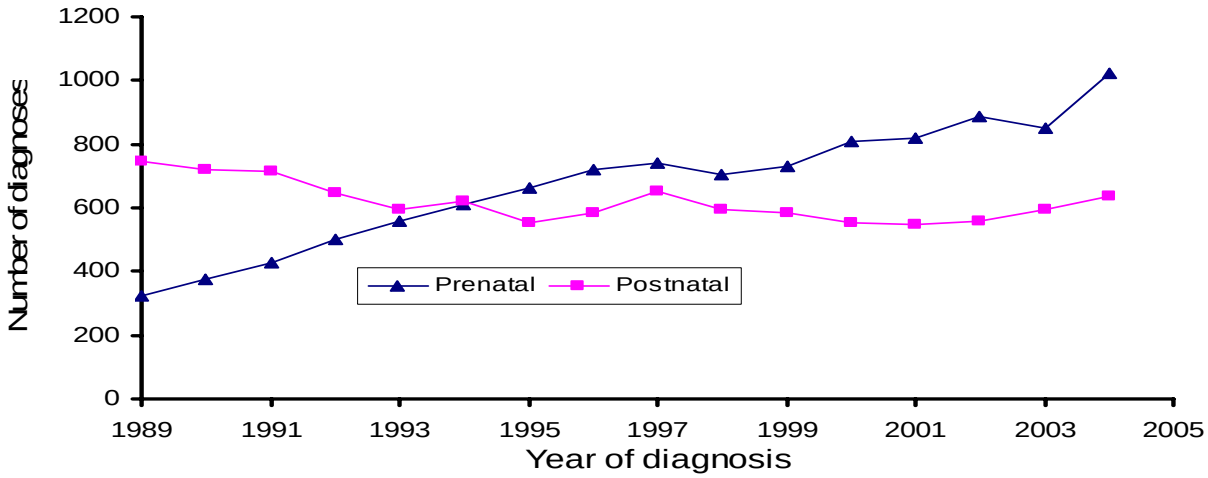
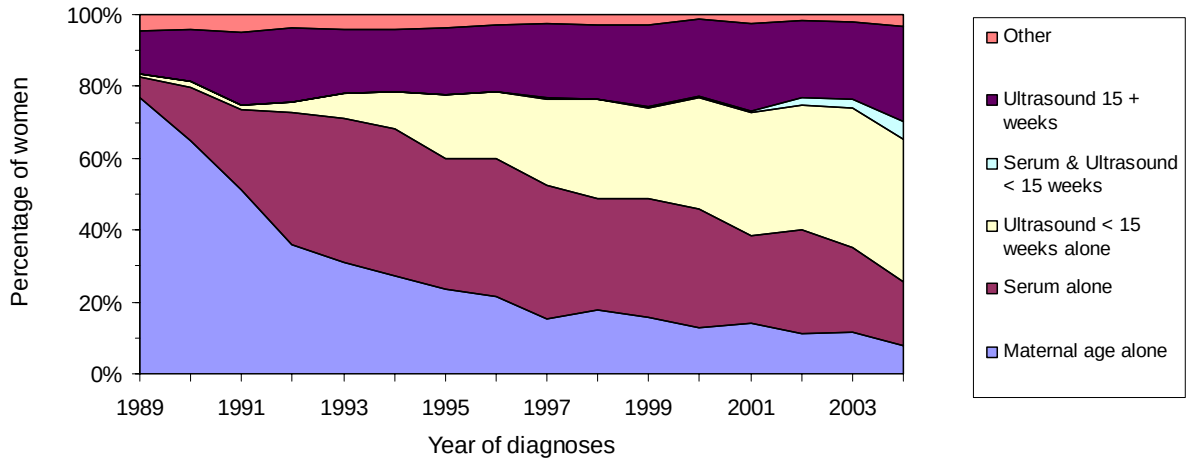


Figure 5: Indication for karyotyping according to year of diagnosis



2004 data are provisional.

Table 6: Down syndrome diagnoses in 2004 according to Government Regional Office (GRO)*

Government Regional Office	No. of diagnoses	% of prenatal diagnoses	% of Indication for Karyotyping					% of tissue sampled	
			Serum	Ultrasound <15 weeks	Serum & Ultrasound <15 weeks	Ultrasound 15+ weeks	Age only reason	CVS	Amnio
North East	75	37	40	16	8	28	8	21	64
North West	150	53	22	17	3	38	18	10	90
Yorkshire and the Humber	123	54	35	12	12	35	5	34	66
East Midlands	114	54	35	19	2	35	5	30	70
West Midlands	149	61	32	18	13	20	10	29	70
East of England	163	65	18	53	3	22	2	58	39
London	337	69	9	61	3	22	3	68	31
South East	242	68	10	55	5	20	9	60	37
South West	124	68	10	34	5	36	13	51	48
Wales	85	65	19	19	0	34	13	15	82
Unknown	97	52	3	35	5	32	16	51	45
Total	1,659	62	18	40	5	27	8	47	51

* 2004 data are provisional.

Table 7: Down syndrome diagnoses and outcomes in England and Wales from 1989 to 2004*

Year	No. diagnoses	% prenatal	No. liveborn	No. TOP	No. Misc[+]/ Still	No. Unknown outcome
1989	1067	30	750	293	16	8
1990	1095	34	738	328	17	12
1991	1144	38	735	369	31	9
1992	1146	44	662	442	24	18
1993	1155	48	622	507	18	8
1994	1234	50	638	542	29	25
1995	1214	54	579	578	32	25
1996	1304	55	606	651	31	16
1997	1390	53	667	658	40	25
1998	1297	54	632	609	21	35
1999	1315	55	606	642	31	36
2000	1365	59	591	686	23	65
2001	1365	60	571	666	30	98
2002	1448	61	590	686	41	131
2003	1444	59	625	635	31	153
2004	1659	62	638	640	58	323
Total	20642	52	10250	8932	473	987

[+] Only miscarriages after prenatal diagnosis are included.
* 2004 data are provisional.

Table 8: Down syndrome prenatal diagnoses 1989 to 2004*

Year	No. of prenatal diagnoses	% of Indication for Karyotyping					Median gestational age (wks)	% of tissue sampled	
		Serum	Ultrasound <15 weeks	Serum & Ultrasound <15 weeks	Ultrasound 15+ weeks	Age only reason		CVS	Amnio
1989	321	6	1	0	12	77	16	18	77
1990	374	15	2	0	14	65	16	16	76
1991	430	22	1	0	20	51	17	15	73
1992	500	37	3	0	21	36	17	11	79
1993	558	40	7	0	18	31	17	17	77
1994	613	41	10	0	17	27	17	23	69
1995	660	37	18	0	18	23	16	25	69
1996	721	38	19	0	19	22	16	30	65
1997	739	37	24	0	21	15	16	35	61
1998	704	31	28	0	21	18	16	35	61
1999	729	33	25	0	23	16	16	33	60
2000	811	33	31	0	21	13	16	37	59
2001	819	25	34	0	24	14	15	45	52
2002	888	29	35	2	21	11	15	43	55
2003	850	24	39	2	22	12	15	46	52
2004*	1021	18	40	5	27	8	15	47	51
Total	10738	30	23	1	21	23	16	33	62

* 2004 data are provisional.

Appendix A

List of Cytogenetic Laboratories in England and Wales

1. Northern Genetics Service
2. Central Manchester and Manchester Children's Hospital
3. Cheshire and Merseyside Genetics Service
4. Yorkshire Regional Genetics Service
5. North Trent Genetics Service
6. Nottingham Genetics Service
7. Leicestershire Genetics Centre
8. West Midlands Regional Genetics Service
9. Oxford Regional Genetics Service
10. East Anglia Regional Genetics Service
11. Norwich Molecular and Cytogenetics Service
12. South Western Regional Genetics Service
13. NW Thames Regional Genetics Service
14. NE Thames Regional Genetics Service
15. SW Thames Regional Genetics Centre
16. Guy's and St Thomas' Hospital NHS Trust
17. Wessex Clinical Genetics and Laboratory Service
18. Cardiff, Wales
19. Cytogenetics Services
20. TDL Genetics (Cytogenetics Services up till 20/02/04)

Appendix B

Data Completeness

The following table gives the completeness of different data items for the years 1989 to 2001, 2002, 2003 and 2004. We are still following up the missing data for 2002 and 2003. The data from 1989 to 2001 are included for comparison purposes to demonstrate the levels of completeness we are aiming to achieve for the 2002, 2003 and 2004 data.

Table B1: Completeness of data from 1989 to 2004*

	Percentage complete			
Data Item	1989-2001	2002	2003	2004*
Reason for referral for karyotyping	100	100	100	99
Type of tissue karyotyped	100	100	100	99
Sex of fetus (some DNA based diagnoses such as FISH and q-PCR do not include sex chromosome analysis)	100	100	98	96
Maternal age	97	92	89	90
Gestational age at sample for prenatal diagnosis	100	100	100	100
Outcome of pregnancy	97	91	89	78
Gestational age at outcome for prenatal diagnosis	84	72	66	60
Number of previous pregnancies	68	58	57	55
Post Codes (some information)	92	96	97	89
(complete postcodes)	84	85	83	82

* 2004 data are provisional.

Appendix C

New form

Reference No. 7234 NDSCR |__|__|__|__|__|

NATIONAL DOWN SYNDROME CYTOGENETIC REGISTER

NOW INCLUDING TRISOMIES 13 AND 18 *(In collaboration with the National Screening Committee)*

LABORATORY: PLEASE RETURN THIS FORM TO: **(Please send blue and green copies to the referring clinician)**

Joan Morris, Wolfson Institute of Preventive Medicine, Barts and the London, Charterhouse Square, London, EC1M 6BQ
Telephone: 0207 882 6220 Fax: 0207 882 6221 Website: http://www.wolfson.qmul.ac.uk/ndscr/

PLEASE ENTER CASE DETAILS AND TICK APPROPRIATE BOXES

Laboratory ID |__|__| PLEASE INCLUDE TRISOMIES 13 & 18

Specimen ID |__|__|__|__|__|__|__|__|__|__|

Karyotype .....

STAGE AT DIAGNOSIS

Prenatal |__| Postnatal |__| After miscarriage |__|

INDICATION FOR KARYOTYPE

Screen +ve |__| Mat age alone |__| Postnatal signs |__|

Other referral reasons: .....

(e.g. anxiety, history, ultrasound markers, etc.) .....

Confirmation of previous diagnosis |__|

ID |__|__|__|__|__|__|__|__|__|__|

SCREENING TEST (both positive and negative results)

Offered |__| Not offered |__|

Accepted |__| Declined |__| >20 weeks |__| other |__|

Risk 1 in ..... Test result: +ve |__| -ve |__|

Please tick all markers measured

1st trimester		2nd trimester	
NT	|__|	AFP	|__|
Papp-A	|__|	uE3	|__|
Free ß-hCG	|__|	hCG (total or freeß)	|__|
		Inhibin-A	|__|

If other combination, please specify.....

CYTOGENETICS

Tissue		Method of karyotyping	
CVS	|__|	Full	|__|
Amniotic Fluid	|__|	PCR	|__|
Fetal Blood	|__|	FISH	|__|
Postnatal Blood	|__|		
Skin	|__|		
Placenta	|__|	Other:	.....

D D M M Y Y

Date sample taken |__|__|__|__|__|__|

Date of mother's LMP |__|__|__|__|__|__|

Gestation when sample taken |__|__| wks

FOR AUDIT PURPOSES PLEASE COMPLETE

Mother's hospital ID |__|__|__|__|__|__|__|__|__|__|

First 3 letters of mother's surname |__|__|__| Initial |__|

Mother's NHS No. |__|__|__|__|__|__|__|__|__|__|

Infant's hospital ID |__|__|__|__|__|__|__|__|__|__|

D D M M Y Y age

Mother's date of birth |__|__|__|__|__|__| |__|__|
(age at testing)

Father's date of birth |__|__|__|__|__|__| |__|__|
(age at testing)

OUTCOME

Termination |__| Miscarriage |__| Stillborn |__| Liveborn |__|

D D M M Y Y

Date of outcome/ termination/ birth |__|__|__|__|__|__|

Gestation at outcome |__|__| wks

Infant's birth weight |__|__|__|__| gms

Please specify consultant and hospital providing pre/postnatal care

.....

If terminated or delivered elsewhere, please specify hospital:

.....

Number of foetuses / babies in this pregnancy |__|

Sex/ outcome of unaffected twin, triplet, etc.

.....

Number of previous pregnancies: (not including this pregnancy)

Termination |__| Miscarriage |__| Stillborn |__| Liveborn |__|

Previous anomalies (number): Trisomy 21 |__| 18 |__| 13 |__|

Other chrom. anomaly (spec.) .....

If previous anomaly, please specify outcome .....

Mother's usual town of residence:

.....

Postcode |__|__|__|__| |__|__|__| PCT |__|__|__|

Note: (e.g. condition of infant, previous family history, fertility problems, etc.)

.....

.....

.....

(Please continue overleaf)

.....

FOR OFFICE USE ONLY

Reference No. 7234 NDSCR |__|__|__|__|__|

Laboratory |__|__| Trisomy 21 18 13 pren/ postn/ misc

Specimen ID |__|__|__|__|__|__|__|__|__|

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 Screen* 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 Obstet Gynaecol* 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 Gynaecol* 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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Appendix E

Useful Websites:

NDSCR
www.wolfson.qmul.ac.uk/ndscr

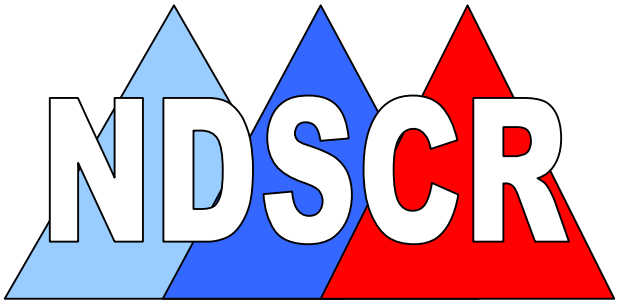
DS Medical Interest Group,
Down syndrome Association, UK
www.dsa-uk.com

Down Syndrome Health Issues
www.ds-health.com

Association of Clinical Cytogeneticists
www.cytogenetics.org.uk

Conclusions

- The NDSCR is approved to gain support under Section 60 of the Health and Social Care Act 2001 and has ethical approval from Trent MREC.
- The NDSCR has continued to maintain a near complete record of all Down syndrome diagnoses in England and Wales in 2004.
- In 2004 there were 1,659 diagnoses of Down syndrome, of which 62% were prenatally diagnosed.
- In 2004 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 outcomes 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.
- Data collection by the NDSCR is funded by the National Screening Committee. The NDSCR is working with the regional and local screening co-ordinators to help them fulfil their audit function.
- 492 Diagnoses of Edwards and Patau syndrome have been included in this report for the first time.



(data collection funded by the National Screening Committee)

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