

National Down Syndrome Cytogenetic Register



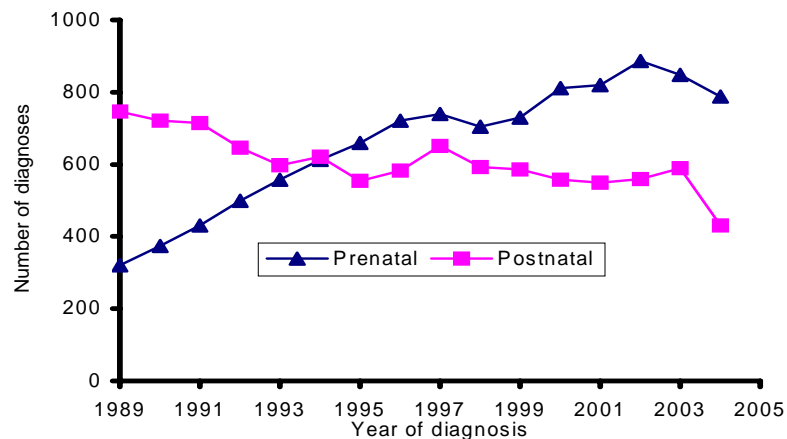
Supplementary Report 2005

The NDSCR started collecting data from the clinical cytogenetic laboratories on all diagnoses involving trisomy 21 over 15 years ago. Thanks to your help the register currently contains 20,000 cases. In this brief report we summarise new developments during the past year and present some preliminary data from 2004 on trisomies 21, 18 and 13.

Aims of the NDSCR

The National Down Syndrome Cytogenetic Register (NDSCR) was established in 1989. The register collects all cytogenetic or DNA reports in England and Wales of trisomies 21, 18 and 13 and their variants. These data are used to 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). The register provides data on annual numbers of affected births to help those planning for their health, educational and social care, and also enables research into the epidemiology of Down, Edwards and Patau syndromes. The data on births is estimated to be over 94% complete.

Figure 1: The number of prenatal and postnatal diagnoses according to year of diagnoses (2004 figures are provisional)



Developments in 2005

1. NDSCR Steering Committee

The following members of the NDSCR steering committee met for the first time in May 2005:

Ms Carol Boys (Chief Executive Office, Down Syndrome Association)
Dr Jenny Kurinczuk (Consultant Clinical Epidemiologist, National Perinatal Epidemiology Unit)
Dr Joan Morris (Director NDSCR - Chair of Steering Committee)
Professor Charles Rodeck (Head of Dept of Obst and Gyn, UCLH London)
Dr Jonathan Waters (Deputy Director, NE London Cytogenetic Laboratory)

The terms of reference of the steering committee are:

To be an independent source for:

- Monitoring the progress of the register towards its overall objectives;
- Advising on the strategies for the use and development of the register;
- Advising on the undertaking and conduct of new research projects;
- Providing technical advice.

2. Membership of EUROCAT

The NDSCR became an affiliate member of EUROCAT (European network of population-based Registries for the epidemiologic surveillance of congenital anomalies). EUROCAT was started in 1979 and 39 registries in 19 countries now survey more than 1 million births per year in Europe. Membership is limited to high quality registries, ascertaining terminations of pregnancy as well as births, and will allow us to collaborate with the other European registers.

3. PIAG approval

The NDSCR re-applied for and was successful in obtaining ethical approval to continue collecting our data (from the Trent MREC and PIAG (Patient Information Advisory Group)) through being a member of BINOCAR (British Isle Network of Congenital Anomaly registers).

4. 2003 Annual Report

Detailed analyses of the data collected in 2003 were published in December 2004. 1437 Down syndrome diagnoses were reported in 2003, with 59% diagnosed prenatally (see Figure 1). A copy is available from the website: <http://www.wolfson.qmul.ac.uk/ndscr>.

5. Trisomies 18 and 13

2004 is the first year the NDSCR added data for trisomies 18 and 13. About 80% of diagnoses received in 2004 were trisomy 21, with 15% being trisomy 18 and 5% trisomy 13 (see Tables 1 and 2).

6. Recent publications in 2004/5

- i. Kovaleva NV, Mutton DE. Epidemiology of double aneuploidies involving chromosome 21 and the sex chromosomes. *Am J Med Genet* 2004; 9999:1-9. ISSN: 01487299.
- ii. Morris JK, de Vigan C, Mutton DE, Alberman E. Risk of a Down syndrome live birth in women of 45 years of age and older. *Prenat Diagn* 2005 Apr;25(4):275-8.

7. Funding

The National Screening Committee has agreed to continue the funding of the register for the years 2005/6.

Table 1: Cases % diagnosed in 2003 and 2004 according to trisomy, time of diagnoses and outcome (2004 figures are provisional)

		2003		2004	
		Trisomy		Trisomy	
		21 No. (%)	21 No. (%)	18 No. (%)	13 No. (%)
Prenatal	Termination	555 (39)	475 (39)	122 (34)	42 (57)
	Livebirth	34 (2)	28 (2)	1 (0)	1 (1)
	Stillbirth/ Miscarriage	15 (1)	17 (1)	8 (4)	3 (4)
	Unknown outcome [†]	244 (17)	282 (23)	73 (32)	23 (31)
Postnatal	Livebirth	575 (40)	403 (33)	21 (9)	5 (7)
	Stillbirth	14 (1)	16 (1)	0 (0)	0 (0)
Total (100%)		1437	1221	225	74

[†] About 8% of the trisomy 21 with unknown outcomes are likely to result in a live birth.

Table 2: Cases diagnosed in 2004 according to trisomy and laboratory

	Trisomy			Total
	21 No. (%)	18 No. (%)	13 No. (%)	No. (100%)
South West	51 (85)	7 (12)	2 (3)	60
Nottingham	31 (82)	5 (13)	2 (5)	38
NE London	83 (77)	22 (20)	3 (3)	108
Manchester	70 (81)	10 (12)	6 (7)	86
Newcastle	31 (97)	1 (3)	0 (0)	32
SE London	59 (87)	5 (7)	4 (6)	68
Wessex	83 (81)	13 (13)	6 (6)	102
Sheffield	44 (94)	3 (6)	0 (0)	47
Leeds	77 (87)	10 (11)	2 (2)	89
Leicestershire	31 (86)	4 (11)	1 (3)	36
SW London	61 (72)	16 (19)	8 (9)	85
Merseyside and Cheshire	24 (77)	6 (19)	1 (3)	31
NW London	67 (71)	20 (21)	7 (7)	94
Birmingham	144 (84)	16 (9)	11 (6)	171
Oxford	29 (73)	11 (27)	0 (0)	40
Cambridge	54 (83)	7 (11)	4 (6)	65
Norwich	8 (80)	2 (20)	0 (0)	10
Cardiff	60 (87)	8 (12)	1 (1)	69
TDL	214 (74)	59 (20)	16 (6)	289
Total	1,221 (80)	225 (15)	74 (5)	1,520

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