The National Down Syndrome Cytogenetic Register

2003 Annual Report

(funded by the National Screening Committee)
Foreword

This 2003 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 2003.

The NDSCR has undergone several changes over the last year:

• This is the first annual report that will include some 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.

• Annabelle Stapleton has joined us to assist Haiyan Wu in running the register.

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
Wayne Huttly
David Mutton
Annabelle Stapleton
Haiyan Wu

December 2004

Contents

The NDSCR

• Introduction
• Aims of the NDSCR
• How the NDSCR works
• What data are collected
• Data completion and processing
• Data confidentiality and informed consent
• How the data are used

The data in the NDSCR

• Down syndrome cases diagnosed in 2003
• Regional differences in cases diagnosed in 2003
• Trends over time in Down syndrome diagnoses

Appendices

Conclusions
Introduction

Welcome to the 2003 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.

The number of Down syndrome pregnancies notified annually has risen from around 1000 in 1989 to 1415 cases in 2003. In November 2003 we first requested notifications of Edwards and Patau syndromes. A total of 35 diagnoses of Patau syndrome and 93 diagnoses of Edwards syndrome were recorded in this first year. These data are not yet complete and we will have to wait for the 2004 data to have an accurate picture of the diagnoses of these syndromes in England and Wales.

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 was made 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

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 by the NDSCR.

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 2000 combined; and separately for 2001, 2002 and 2003. 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 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 the NDSCR as a part of the British Isles Network of Congenital Anomaly Registers (BINOCAR) was given permission to operate without informed consent. This permission was successfully re-applied for in 2004. In 2004 the NDSCR as part of BINOCAR also applied to Trent multi-centre research ethics committee (MREC) for ethical approval, which was granted.

How the data are used

Audit of Down Syndrome Screening

- All local screening co-ordinators should receive the green copy of the NDSCR form which will 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) By combining data from the NDSCR and data from registries who are members of EUROCAT (European Concerted Action on Congenital Anomalies and Twins) (Morris et al, in press) we demonstrated that the age related risk of a Down syndrome pregnancy does not continue increasing over the age of 45 years.

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 risks.

Collaborative studies

1) David Neasham from SASHU (Small Area Health Statistics Unit) has used relevant NDSCR data for his PhD thesis which shows that 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; University of London, 2003).

2) 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.

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, Great Ormond Street Hospital for Sick Children, 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.

The Data in the NDSCR

Down syndrome cases diagnosed in 2003

There were 1415 Down syndrome diagnoses made in 2003, 831 (59%) made prenatally and 584 (41%) postnatally (Table 1 and Figure 1). If we assume that of the prenatal diagnoses with unknown outcomes the proportion terminated remains as before 2003, the likely number of Down syndrome live births in England and Wales in 2003 would have been 624 (28 + 570 + 8% of 324), a birth prevalence of 1 per 1000 livebirths.

Table 1: Down syndrome cases diagnosed in 2003* by time of diagnoses and outcome

<table>
<thead>
<tr>
<th></th>
<th>No.</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Termination of pregnancy</td>
<td>466</td>
<td>33</td>
</tr>
<tr>
<td>Live Birth</td>
<td>28</td>
<td>2</td>
</tr>
<tr>
<td>Still Birth / Miscarriage</td>
<td>13</td>
<td>1</td>
</tr>
<tr>
<td>Unknown outcome†</td>
<td>324</td>
<td>23</td>
</tr>
<tr>
<td>Postnatal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Live Birth</td>
<td>570</td>
<td>40</td>
</tr>
<tr>
<td>Still Birth</td>
<td>14</td>
<td>1</td>
</tr>
<tr>
<td>Total</td>
<td>1415</td>
<td>100</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.
† About 8% of those with unknown outcomes are likely to result in a live birth.
**Indication for prenatal karyotyping**

The indications for karyotyping reflect the different methods of prenatal screening occurring. In 62% of all prenatally diagnosed cases the indication mentioned was the result of an ultrasound scan. 39% of these were carried out before 15 weeks, and were probably nuchal translucency measurements. Unfortunately we cannot determine this precisely from the form used in 2003. This information should be available for 2004 from the revised form. In 25% of prenatally diagnosed cases the indication was a maternal serum test, in 11% 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 831 prenatally diagnosed cases, 25% were diagnosed before 13 weeks, 72% 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.

**Tissue used for karyotyping**

The most common source of sampling fetal cells in 2003 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 9 days for amniocentesis. 91% of all terminations following CVS were within 14 days of the procedure compared with 74% for amniocentesis.
Patau and Edwards syndrome cases diagnosed in the latter part of 2003

The data on Patau and Edwards syndrome are from a few laboratories from November 2003. Around 90% of both the Patau and Edwards syndrome diagnoses were made prenatally (Table 4).

Regional differences in Down syndrome cases diagnosed in 2003

Tables 5 and 6 show the patterns of diagnoses of Down syndrome across England and Wales. The proportion of cases diagnosed prenatally varies from 47% in North West NHS Region to 76% in Wales. The completeness of the data also varies by region with only 9% of outcomes being unknown in Trent compared to 34% in London. 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.

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Patau</th>
<th>Edwards</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal Termination of pregnancy</td>
<td>23</td>
<td>46</td>
</tr>
<tr>
<td>Live Birth</td>
<td>0</td>
<td>4</td>
</tr>
<tr>
<td>Still Birth / Miscarriage</td>
<td>0</td>
<td>6</td>
</tr>
<tr>
<td>Unknown outcome</td>
<td>8</td>
<td>31</td>
</tr>
<tr>
<td>Postnatal Live Birth</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>Still Birth</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Total</td>
<td>35</td>
<td>93</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.

Table 5: Down syndrome diagnoses by NHS Regional Office Area in 2003*

<table>
<thead>
<tr>
<th>Region</th>
<th>No. of diagnoses</th>
<th>% of prenatal</th>
<th>Median maternal age (years)</th>
<th>% of missing maternal age*</th>
<th>% of Unknown outcome*</th>
</tr>
</thead>
<tbody>
<tr>
<td>North &amp; Yorkshire</td>
<td>138</td>
<td>53</td>
<td>36.1</td>
<td>12</td>
<td>18</td>
</tr>
<tr>
<td>Trent</td>
<td>127</td>
<td>63</td>
<td>36.4</td>
<td>3</td>
<td>9</td>
</tr>
<tr>
<td>West Midlands</td>
<td>112</td>
<td>53</td>
<td>37.6</td>
<td>22</td>
<td>10</td>
</tr>
<tr>
<td>North West</td>
<td>171</td>
<td>47</td>
<td>36.3</td>
<td>24</td>
<td>28</td>
</tr>
<tr>
<td>Eastern</td>
<td>140</td>
<td>65</td>
<td>37.3</td>
<td>13</td>
<td>26</td>
</tr>
<tr>
<td>London</td>
<td>317</td>
<td>63</td>
<td>37.4</td>
<td>13</td>
<td>34</td>
</tr>
<tr>
<td>South East</td>
<td>230</td>
<td>57</td>
<td>36.5</td>
<td>7</td>
<td>22</td>
</tr>
<tr>
<td>South West</td>
<td>121</td>
<td>60</td>
<td>37.6</td>
<td>7</td>
<td>19</td>
</tr>
<tr>
<td>Wales</td>
<td>59</td>
<td>76</td>
<td>38.2</td>
<td>5</td>
<td>17</td>
</tr>
<tr>
<td>Total</td>
<td>1415</td>
<td>59</td>
<td>36.9</td>
<td>12</td>
<td>23</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.
+ Data currently being updated.

Table 6: Prenatal diagnoses by NHS Regional Office Area, indication and tissue used for diagnoses in 2003*

<table>
<thead>
<tr>
<th>Region</th>
<th>No. of prenatal diagnoses</th>
<th>% of indication for Karyotyping*</th>
<th>Median gestational age (wks)</th>
<th>% of tissue sampled</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Ultrasound scan</td>
<td></td>
<td>Serum Age</td>
<td>CVS</td>
</tr>
<tr>
<td></td>
<td>&lt; 15 wks</td>
<td>≥ 15 wks</td>
<td>%</td>
<td>Wks</td>
</tr>
<tr>
<td>North &amp; Yorkshire</td>
<td>73</td>
<td>30</td>
<td>34</td>
<td>36</td>
</tr>
<tr>
<td>Trent</td>
<td>80</td>
<td>13</td>
<td>21</td>
<td>41</td>
</tr>
<tr>
<td>West Midlands</td>
<td>59</td>
<td>24</td>
<td>34</td>
<td>41</td>
</tr>
<tr>
<td>North West</td>
<td>81</td>
<td>19</td>
<td>25</td>
<td>22</td>
</tr>
<tr>
<td>Eastern</td>
<td>91</td>
<td>40</td>
<td>21</td>
<td>31</td>
</tr>
<tr>
<td>London</td>
<td>199</td>
<td>62</td>
<td>19</td>
<td>11</td>
</tr>
<tr>
<td>South East</td>
<td>130</td>
<td>57</td>
<td>14</td>
<td>18</td>
</tr>
<tr>
<td>South West</td>
<td>73</td>
<td>29</td>
<td>22</td>
<td>19</td>
</tr>
<tr>
<td>Wales</td>
<td>45</td>
<td>22</td>
<td>33</td>
<td>36</td>
</tr>
<tr>
<td>Total</td>
<td>831</td>
<td>39</td>
<td>23</td>
<td>25</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.
+ More than one indication may be mentioned.
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 59% in 2003, and numbers detected prenatally from 321 to 831 in 2003 (Table 7 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 to the increase in earlier prenatal diagnoses and subsequent terminations. Many of the 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 8). This proportion then decreased with the introduction of nuchal translucency measurements as a screening test. In 2003 a serum test was mentioned as an indication for prenatal diagnosis in 25%, 39% mentioning ultrasound results before 15 weeks gestation as an indication. The use of maternal age alone as an indication for karyotyping is decreasing steadily, although even in 2003 it was given as an indication in 11% 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 46% to 52% respectively in 2003 (Table 8).

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

![Figure 4](image1.png)

Figure 5: Indication for karyotyping according to year of diagnosis
(may exceed 100% due to more than one indication being given)

![Figure 5](image2.png)

2003 data are provisional.
Table 7: Down syndrome diagnoses and outcomes in England and Wales from 1989 to 2003*

<table>
<thead>
<tr>
<th>Year</th>
<th>No. diagnoses</th>
<th>% prenatal</th>
<th>No. liveborn</th>
<th>No. TOP</th>
<th>No. Misc* / Still</th>
<th>No. Unknown outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>1989</td>
<td>1067</td>
<td>30</td>
<td>750</td>
<td>293</td>
<td>16</td>
<td>8</td>
</tr>
<tr>
<td>1990</td>
<td>1095</td>
<td>34</td>
<td>738</td>
<td>328</td>
<td>17</td>
<td>12</td>
</tr>
<tr>
<td>1991</td>
<td>1144</td>
<td>38</td>
<td>735</td>
<td>369</td>
<td>31</td>
<td>9</td>
</tr>
<tr>
<td>1992</td>
<td>1146</td>
<td>44</td>
<td>662</td>
<td>442</td>
<td>24</td>
<td>18</td>
</tr>
<tr>
<td>1993</td>
<td>1155</td>
<td>48</td>
<td>622</td>
<td>507</td>
<td>18</td>
<td>8</td>
</tr>
<tr>
<td>1994</td>
<td>1234</td>
<td>50</td>
<td>638</td>
<td>542</td>
<td>29</td>
<td>25</td>
</tr>
<tr>
<td>1995</td>
<td>1214</td>
<td>54</td>
<td>579</td>
<td>578</td>
<td>32</td>
<td>25</td>
</tr>
<tr>
<td>1996</td>
<td>1304</td>
<td>55</td>
<td>606</td>
<td>651</td>
<td>31</td>
<td>16</td>
</tr>
<tr>
<td>1997</td>
<td>1390</td>
<td>53</td>
<td>667</td>
<td>658</td>
<td>40</td>
<td>25</td>
</tr>
<tr>
<td>1998</td>
<td>1297</td>
<td>54</td>
<td>632</td>
<td>609</td>
<td>21</td>
<td>35</td>
</tr>
<tr>
<td>1999</td>
<td>1315</td>
<td>55</td>
<td>606</td>
<td>642</td>
<td>31</td>
<td>36</td>
</tr>
<tr>
<td>2000</td>
<td>1370</td>
<td>59</td>
<td>592</td>
<td>677</td>
<td>23</td>
<td>78</td>
</tr>
<tr>
<td>2001</td>
<td>1368</td>
<td>60</td>
<td>570</td>
<td>568</td>
<td>28</td>
<td>202</td>
</tr>
<tr>
<td>2002</td>
<td>1446</td>
<td>61</td>
<td>585</td>
<td>534</td>
<td>38</td>
<td>289</td>
</tr>
<tr>
<td>2003*</td>
<td>1415</td>
<td>59</td>
<td>598</td>
<td>466</td>
<td>27</td>
<td>324</td>
</tr>
<tr>
<td>Total</td>
<td>18960</td>
<td>51</td>
<td>9580</td>
<td>7864</td>
<td>406</td>
<td>1110</td>
</tr>
</tbody>
</table>

+ Only miscarriages after prenatal diagnosis are included.
* 2003 data are provisional.

Table 8: Down syndrome prenatal diagnoses 1989 to 2003*

<table>
<thead>
<tr>
<th>Year</th>
<th>No. of prenatal diagnoses</th>
<th>% of Indication for Karyotyping*</th>
<th>Median gestational age (wks)</th>
<th>% of tissue sampled</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Ultrasound scan</td>
<td></td>
<td>CVS</td>
</tr>
<tr>
<td></td>
<td></td>
<td>&lt;15 wks</td>
<td>≥15 wks</td>
<td>Serum</td>
</tr>
<tr>
<td>1989</td>
<td>321</td>
<td>1</td>
<td>12</td>
<td>6</td>
</tr>
<tr>
<td>1990</td>
<td>374</td>
<td>2</td>
<td>15</td>
<td>15</td>
</tr>
<tr>
<td>1991</td>
<td>430</td>
<td>1</td>
<td>21</td>
<td>22</td>
</tr>
<tr>
<td>1992</td>
<td>500</td>
<td>3</td>
<td>24</td>
<td>37</td>
</tr>
<tr>
<td>1993</td>
<td>558</td>
<td>7</td>
<td>21</td>
<td>40</td>
</tr>
<tr>
<td>1994</td>
<td>613</td>
<td>11</td>
<td>19</td>
<td>41</td>
</tr>
<tr>
<td>1995</td>
<td>660</td>
<td>18</td>
<td>22</td>
<td>37</td>
</tr>
<tr>
<td>1996</td>
<td>721</td>
<td>20</td>
<td>21</td>
<td>38</td>
</tr>
<tr>
<td>1997</td>
<td>739</td>
<td>25</td>
<td>24</td>
<td>38</td>
</tr>
<tr>
<td>1998</td>
<td>704</td>
<td>29</td>
<td>22</td>
<td>31</td>
</tr>
<tr>
<td>1999</td>
<td>729</td>
<td>26</td>
<td>25</td>
<td>34</td>
</tr>
<tr>
<td>2000</td>
<td>812</td>
<td>32</td>
<td>24</td>
<td>33</td>
</tr>
<tr>
<td>2001</td>
<td>819</td>
<td>36</td>
<td>25</td>
<td>26</td>
</tr>
<tr>
<td>2002</td>
<td>886</td>
<td>37</td>
<td>23</td>
<td>31</td>
</tr>
<tr>
<td>2003*</td>
<td>831</td>
<td>39</td>
<td>23</td>
<td>25</td>
</tr>
<tr>
<td>Total</td>
<td>9697</td>
<td>23</td>
<td>22</td>
<td>31</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.
* 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 6533

ROYAL MANCHESTER CHILDREN’S HOSPITAL
Hospital Road, Pendlebury, Manchester, M27 4HA
Tel: 0161 727 2567 (now merged with the Regional
Genetic Service based at St Mary’s Hospital in
Manchester)

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 C

Data Completeness

The following table gives the completeness of different data items for the years 1989 to 2000, 2001, 2002 and 2003. We are still following up the missing data for 2002 and 2003. The data from 1989 to 2000 are included for comparison purposes to demonstrate the levels of completeness we are aiming to achieve for the 2001, 2002 and 2003 data.

Table C1: Completeness of data from 1989 to 2003*

<table>
<thead>
<tr>
<th>Data Item</th>
<th>1989-2000</th>
<th>2001</th>
<th>2002</th>
<th>2003*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reason for referral for karyotyping</td>
<td>100</td>
<td>100</td>
<td>99</td>
<td>100</td>
</tr>
<tr>
<td>Type of tissue karyotyped</td>
<td>100</td>
<td>99</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Sex of fetus (some DNA based diagnoses such as FISH and q-PCR do not include sex chromosome analysis)</td>
<td>100</td>
<td>100</td>
<td>98</td>
<td>98</td>
</tr>
<tr>
<td>Maternal age</td>
<td>97</td>
<td>92</td>
<td>91</td>
<td>89</td>
</tr>
<tr>
<td>Gestational age at sample for prenatal diagnosis</td>
<td>100</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Outcome of pregnancy</td>
<td>97</td>
<td>85</td>
<td>80</td>
<td>77</td>
</tr>
<tr>
<td>Gestational age at outcome for prenatal diagnosis</td>
<td>84</td>
<td>66</td>
<td>61</td>
<td>56</td>
</tr>
<tr>
<td>Number of previous pregnancies</td>
<td>66</td>
<td>58</td>
<td>52</td>
<td>52</td>
</tr>
<tr>
<td>Post Codes (some information) (complete postcodes)</td>
<td>92</td>
<td>94</td>
<td>95</td>
<td>90</td>
</tr>
</tbody>
</table>

* 2003 data are provisional.
Appendix D

NDSCR Publications


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 2003.
- In 2003 there were 1,415 diagnoses of Down syndrome, of which 59% were prenatally diagnosed.
- In 2003 the Down syndrome live birth rate was around 1 per 1000 (this figure is provisional as there are a large number of missing outcomes).
- 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.
- The NDSCR is funded by the National Screening Committee and is working with the regional and local screening co-ordinators to help them fulfil their audit function.
- Some diagnoses of Edwards and Patau syndrome diagnoses have been included in this report for the first time.
Wolfson Institute of Preventive Medicine
Centre for Preventive and Environmental 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 6274/ 6217/ 6220
Fax: 020 7882 6221
Email: ndscr@qmul.ac.uk