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*Antenatal and Newborn
Screening Programmes*

National Down's Syndrome
Screening Programme for England

3rd Annual Programme Report
2005/2006

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Abbreviations

CfH	Connecting for Health
CNST	Clinical Negligence Scheme for Trusts
CPA	Clinical Pathology Accreditation
CVS	Chorionic Villus Sampling
DIPEX	Database Index of Patients' Experiences
DQASS	Down's Syndrome Quality Assurance Support Service
DR	Detection Rate
FMCH	Fetal and Maternal Child Health sub group
FPR	False Positive Rate
HTA	Health Technology Assessment
MHRA	Medicines and Healthcare products Regulatory Agency
NDSCR	National Down's Syndrome Cytogenetic Register
NEQAS	National External Quality Assessment Service
NPfIT	National Programmes for Information Technology
NT	Nuchal Translucency
QA	Quality Assurance
ROCR	Review of Central Returns
SPR	Screen Positive Rate
UKNSC	UK National Screening Committee

Preface

Welcome to the third annual report of the NHS Down's syndrome Screening Programme. The format of the report has been changed to become cohesive with the other national screening programmes. The corporate image is reflected in the new logo of the Antenatal and Newborn Screening Programmes however the Down's syndrome Screening Programme continues to use its yellow colour as an identifiable corporate image.

Statistics and information produced will concord with the agreement of the UK National Screening Committee (UKNSC) directorate that we should produce information in relation to a fiscal year. This report incorporates information on national figures to reflect our commencement on the quality assurance and performance management of the programme as a whole. It is expected that statistical information will be extended in the future as data collection systems begin to align and function.

The report outlines the progress and work produced during 2005 / March 2006 and relates the progress to the programme objectives. The key aim being to implement Down's syndrome screening in all maternity hospitals. This has been achieved in all but one.

The National Programme continues to work under the umbrella of the UK National Screening Committee which itself has undergone changes to its group structure.

The UKNSC now has one sub group for antenatal and neonatal screening. This is called the Fetal, Maternal and Child Health group (FMCH) which replaces the two separate Antenatal and Child Health sub groups of the UK National Screening Committee. The terms and remit of the UKNSC have not changed although the work has become more extensive and cross cutting in relation to the full implementation of the antenatal and newborn screening programmes.

During 2006 there will be changes to the relationships and structures with whom the National Programme works. This is particularly so in respect of the public health departments who have undergone further restructuring arrangements. It is not expected that the structure of the National Screening Programmes will alter substantially during this restructuring but continue to act as a stable support to those public health departments whilst they develop their new roles.

Overall the National Programme has achieved its objectives that were set out in the last report and will continue to work towards a uniform national screening programme for England.

The National Programme continues to be assisted by many experts and professionals in the field and without their valued support and knowledge we would not be able to improve services for women.

P A Ward

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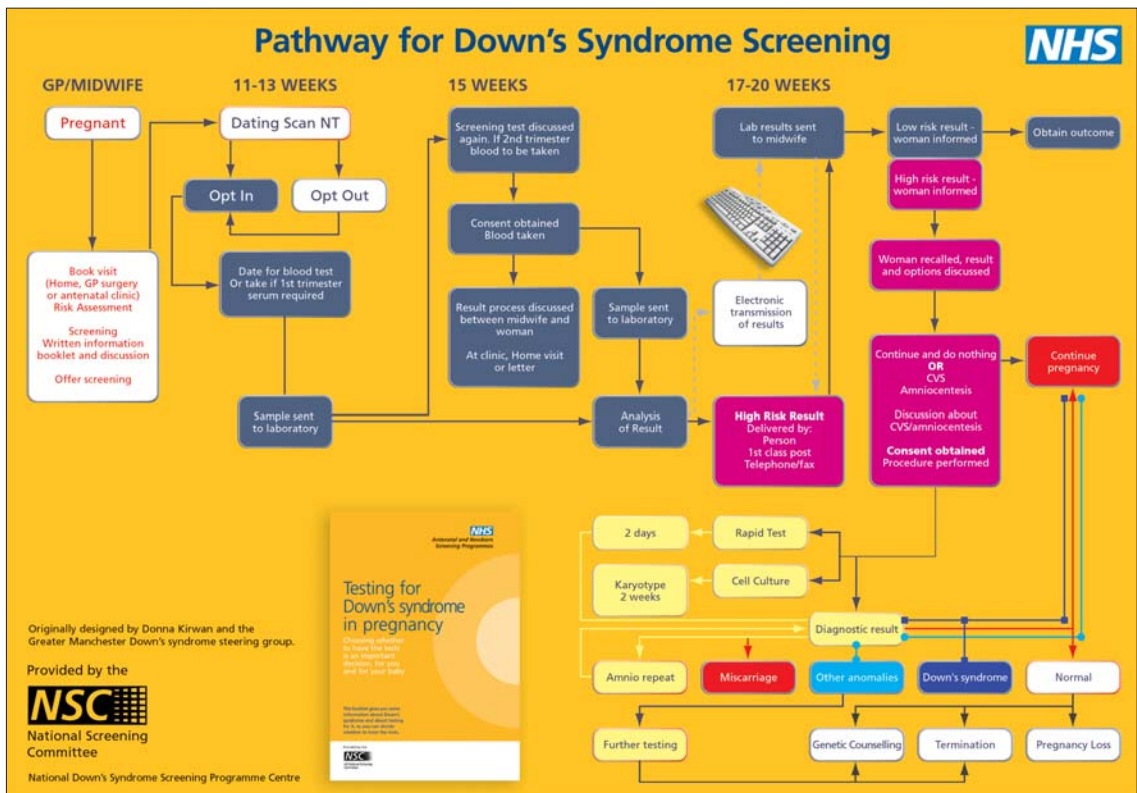
1.0 The Aim and Objectives of the programme

The main aim and objective of the programme remains the same, which is to ensure access to a uniform screening programme which conforms to an agreed level of quality for all pregnant women in England who undergo screening for Down's syndrome. This is supported by key aims to provide information for women so that they are able to exercise informed choice.

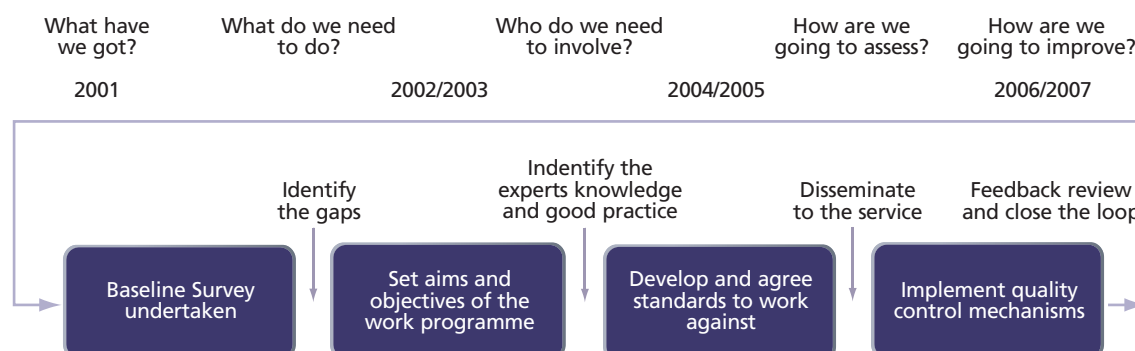
During 2005/2006 we have begun to establish firm quality assurance mechanisms to improve the testing strategies in place as well as achieving full implementation in all but one Trust. Ensuring the quality of the test result is a key objective of the future programme.

The screening programme encompasses the diagnostic pathway in this particular programme and efforts have been made to clarify the preferred diagnostic method that should be offered to women. This is particularly so around the discussion and implementation of rapid diagnostic techniques that should now be available to all women undergoing screening.

The screening and diagnostic pathway can be seen in diagram 1



The pathway to secure a quality screening programme which has been followed since 2001 is set out below in diagram 2.



2.0 Provenance of the Programme

The National Programme is part of the overall structure of the UK National Screening Programmes Directorate. This is overseen by the UK National Screening Committee which was set up by Ministers in 1996. The Director of the UKNSC is Sir Muir Gray.

The terms and remit of the UKNSC can be seen below. <http://www.nsc.nhs.uk/>

The UK National Screening Committee (NSC) advises Ministers, the devolved National Assemblies and the Scottish Parliament on all aspects of screening policy. It has a Fetal Maternal and Child Health Co-ordinating Group (FMCH) which deals with antenatal and child health screening issues. In forming its proposals, the NSC draws on the latest research evidence and the skills of specially convened multi-disciplinary expert groups, which always include patient and service user representatives.

The NSC assesses proposed new screening programmes against a set of internationally recognised criteria covering the condition, the test, the treatment options and effectiveness and acceptability of the screening programme. Assessing programmes in this way is intended to ensure that they do more good than harm at a reasonable cost. In 1996, the NHS was instructed not to introduce any new screening programmes until the NSC had reviewed their effectiveness. This was communicated in EL96 (110).

The Remit and Terms of Reference of the UK National Screening Committee:

The remit and terms of reference of the National Screening Committee are:

- (i) The UK National Screening Committee will advise Ministers, the devolved National Assemblies and the Scottish Parliament on:
- the case for implementing new population screening programmes not presently purchased by the NHS within each of the countries in the UK;
 - screening technologies of proven effectiveness but which require controlled and well-managed introduction;
 - the case for continuing, modifying or withdrawing existing population screening programmes. In particular, programmes inadequately evaluated or of doubtful effectiveness, quality or value.

- (ii) The NSC will call on sound evidence to inform its advice and recommendations. In particular:
 - (a) calling on the advice of the Standing Group on Health Technologies Diagnostic Panel (formerly the Population Screening Panel) and in turn inform the setting of NHS R&D priorities;
 - (b) calling on the DH Policy Research Programme and defining research needs for screening;
 - (c) calling on other and appropriate sources of sound evidence from within and outside the NHS.
- (iii) The NSC will set up practical mechanisms to oversee the introduction of a new programme and its implementation in the NHS. It will monitor effectiveness and quality assurance.
- (iv) The NSC will be informed by reports from the Advisory Groups for specific programmes on the performance of those programmes, and issues that arise which would have relevance to general screening policy.

3.0 Governance of the Programme

The Down's syndrome national screening programme is one of a number of nationally managed programmes.

The Director of the Down's syndrome National Programme, Mrs. Pat Ward, is responsible to the overall Programmes Director, Sir Muir Gray, Director of the UK National Screening Committee. The Programme Director and staff working within the National Programme Centre are employed by the Oxford Radcliff Hospitals Trust. The base office is known as the Programme Centre.

4.0 Supporting Staff and Groups

4.1 Supporting staff

The Programme Centre is supported by a number of contracted staff who also oversee the National Fetal Anomaly Ultrasound Programme and has changed slightly since the last report:-

- 1 wte programme director.
- 1.6 wte secretarial support (increased since 2004).
- 1 wte midwifery assistance (increased since 2004).
- 0.2 wte consultant biochemist.
- 0.2 wte consultant radiographer.
- 0.4 wte statisticians (new since 2004).
- 3 consultants in an honourarium capacity (one new consultant since 2004).

The national antenatal training and education lead is funded and accountable to the Programme Centre on a 0.8 wte basis (increased since 2004).

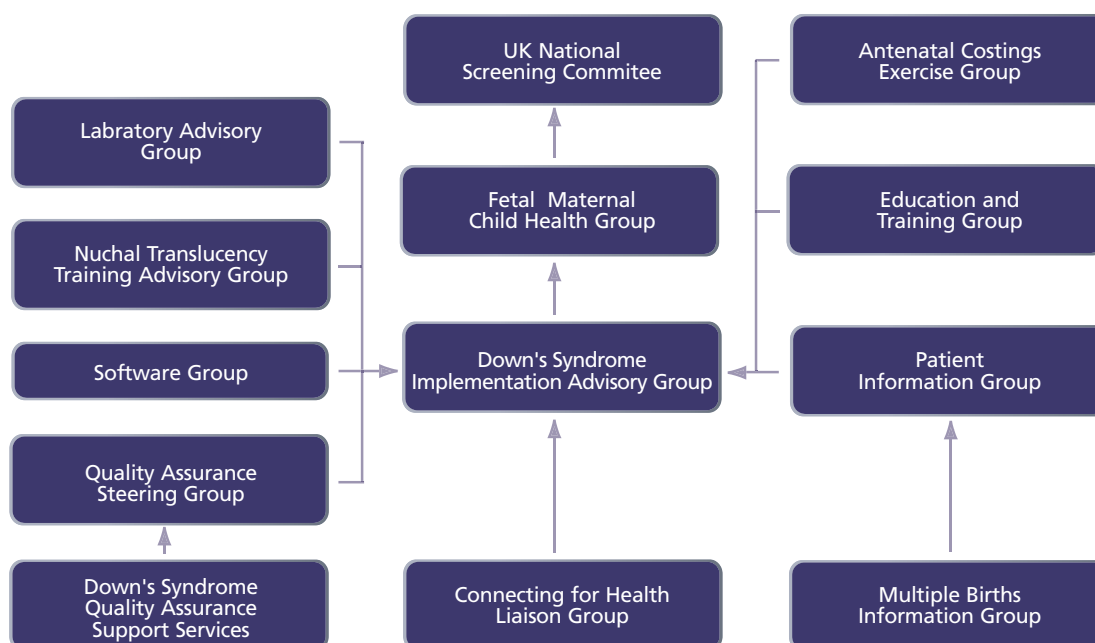
4.2 Working groups

A number of working groups support the programme with their expert knowledge and advice. The overarching group is the Down's syndrome implementation advisory group and this is chaired by Professor Peter Soothill. One extra group that has been formed during 2005/6 has been the Quality Assurance group. The remit of this group is to take forward quality issues, initially for the test itself and then expanding this to the rest of the programme.

We have also prepared information about screening in multiple pregnancies as well as

re-establishing the audit group which is now known as the Connecting for Health Liaison group. Membership of the groups can be seen in appendix 1. A diagram of the group structure can be seen below. Further information about the work and output of these groups is documented in this report.

Diagram 3. The group structure of the National Programme



5.0 Programme Development and Management activities

Overview of Activities during 2005/6

Apart from the continuing activities of the work of the groups a number of initiatives have taken place. These are:-

5.1 Initiatives

- Funding and developing the DQASS system which will underpin the quality of the test including the basis of a national database.
- Continual working with commercial suppliers and setting a specification to improve the service they provide to the National Programme.
- Funding of the National Down's syndrome Cytogenetic Register.
- A continuation of the funding for training for Nuchal Translucency measurement in England.
- Funding and completion of the work of patients' experiences through the DIPEX web site. <http://www.dipex.org/antenatalscreening> 500 CDs have been bought and distributed to the service.
- Developing and funding of a gestational calculator wheel for midwives to use. 5000, distributed to the service.
- Part developing and funding of resource cards for midwives. <http://www.screening.nhs.uk/cpd/home.htm>

- Funding, implementation and evaluation of the education and training pack into the service.
- Funding of training days for biochemists in Down's syndrome screening.
- The production of quarterly newsletters to service staff.
- Commissioning Leeds University to assess ultrasound machinery used for NT measurement, with the aim of producing a specification for the service to work to.
- Undertaking a complete review of the standards which support the programme.
- Funding a pilot site for the contingency screening strategy in the West Midlands which will report next year.
- Continual working with Trusts to achieve full implementation of the Down's syndrome screening programme.
- Development of patient information for multiple pregnancies
- Development of patient information for diagnostic testing.
- Printing of a further 250,000 patient information leaflets and the distribution of those.
- Funding of further 9 training days for sonographers by ARC (Antenatal Results and Choices).
- A review of the present overall screening policy. This work is still in progress.

6.0 Implementation of the National Screening Programme in relation to the key aim and objectives.

The original key aim of the programme was to fulfil the commitment given by the Minister for Health in 2001 that all pregnant women would be offered screening for Down's syndrome. Working with the NHS service this has now almost been achieved.

In the original survey of 2001, 44 of Trusts (25%) offered either no screening or a restricted service, to their pregnant population. At the end of 2004 there were 6 Trusts who had not implemented screening for Down's syndrome for their pregnant population. Further work and discussion with the Regional Antenatal Screening Teams for England has ensured that all Trusts, except one, have implemented screening to all of their pregnant population by April 2006. All but 2 Trusts have dating scan provision prior to a screening test taking place. One of the standards of the programme is that all screening for Down's syndrome must be preceded by an ultrasound scan to assess gestational age. Clearly those Trusts who do not have this in place are outside of the standard set by the programme and those Trusts are being informed of our concerns about possible litigation.

In 2002 dating scan provision was only offered by 87% of those offering screening which equated to 70%, (146/208) of the pregnant population being able to access a dating scan. Recognition has to be given to the achievement of implementing a dating scan service for almost all women during a period of scarce ultrasound resources and finances.

The majority of screening, around 70% is still second trimester biochemical screening using the triple test. One of the aims of the Model of Best Practice 2003 policy (DH) is to ensure that a minimum of a triple test is in place by 2005. The achievement of this has been difficult but is now almost complete. Since the original survey in 2001 the service has moved from 75% of screening for women to 99.9%. No extra money has been allocated for this implementation and Trusts have provided the extra services in an arena of financial difficulties within the NHS. The differing screening strategies in place as of April 2006 can be seen in table 1.

Table 1:

Present strategies used	Number of hospitals	Correlating to region	Total
Double Test	8,7,6,2,	SW,NW,EM,WM	23
NT Only	3,1,1,13,11	SW,NE,E,EM,SE,L	29
Combined	1,8,6,4	SW,NW,SE,L	19
Triple Test	4,26,17,16,19,9,15,11,7	SW,NW,NE,E,Y+H,EM,WM,SE,L	124
Quadruple	4,1,4,3	NW,E,SE,L	12
Integrated	2	L	2
Serum Integrated	3	SW	3
Total			212

There have been, and remain, difficulties in implementing 1st trimester nuchal translucency (NT) services despite the offer of free NT training from the National Programme. The National Programme is looking at ways to improve capacity in the ultrasound departments as part of the work of the National Fetal Anomaly Ultrasound Screening programme which is managed from the same office. This is explained more fully in section 8.4.3.

A concern has been the large number of laboratories that undertake analysis of the test which in turn created a small sample throughput in those laboratories. One of the standards set by the expert group to the National Programme is that to achieve an adequate assessment of the efficacy and quality of the screening test then a minimum sample throughput was set at 5,000 for a laboratory. In 2001, 55 laboratories undertook screening. The objective for the programme has been to reduce the number of laboratories through discussions and reconfiguration of the service to achieve that standard. In 2005/2006 the number of laboratories has been reduced from 55 to 30. This may be further reduced in the future as new standards are being set to increase sample throughput. The rationale is that the confidence intervals can be tighter with a higher sample throughput and hence the stated detection and false positive rates will be more accurate.

Assessment of the biochemical testing has been relatively weak in the past and this has been acknowledged. Although there is an external quality assurance scheme (NEQAS), this has been set up on an educational base and has no authoritative role. The National Programme has addressed this by setting up an external QA mechanism and 10 laboratories of the 30 have taken part in this scheme. Further work in this area will continue as quality control mechanisms are developed to support the aim of improving the quality of the test.

Other objectives are to put into place an audit and monitoring system so that assessment of overall programme efficacy can take place. The audit objective will deliver against all of the programme standards and some of the data collection points can be seen in the next section. This will be a future development objective for 2006/2007.

A key objective has been to produce information for women that is uniform and accessible. The standard that supports this is that all women should be given information prior to their screening test with adequate time to make a decision about the offer of screening. The national leaflet has been distributed to all hospitals and in the latest survey was given out by 74% of them.

Education of staff is an important area to ensure that the screening test is delivered to a set standard. Standards around education and training have been set out, and to ensure that they can be met, the National Programme has developed and disseminated a training package to all midwifery staff. This has been delivered by the Regional Educational and Training teams funded by the NSC. The reviews of this resource and its effect have been very good.

7.0 Statistics for 2005/6

7.1 Collection Points

There are a number of data collection points which are beginning to produce figures for the programme. It is still in an embryonic state, and the full deliverance of all data required for the programme is not expected to be fully in place until Connecting for Health (CfH, the new IT system for all England), has delivered against our specification. We have been informed that this may not be until at least 2008 and possibly 2010. Until then the programme is reliant on other methods to inform us of progress. However we do fund 2 specific areas where data is being delivered to the programme. This is through the National Down's syndrome Cytogenetic Register (NDSCR) and the Down's syndrome Quality Assurance Support service (DQASS) as well as small on going audits of all Trusts. Data points and definitions have been put forward to the Review of Central Returns (ROCR) for collection at an all Trust level. It is expected that this will be accepted by ROCR with basic information such as information on uptake, detection and false positive rates being delivered to the programme in 2008. <http://www.ic.nhs.uk/rocr>

7.2 NDSCR (The National Down's Syndrome Cytogenetic Register)

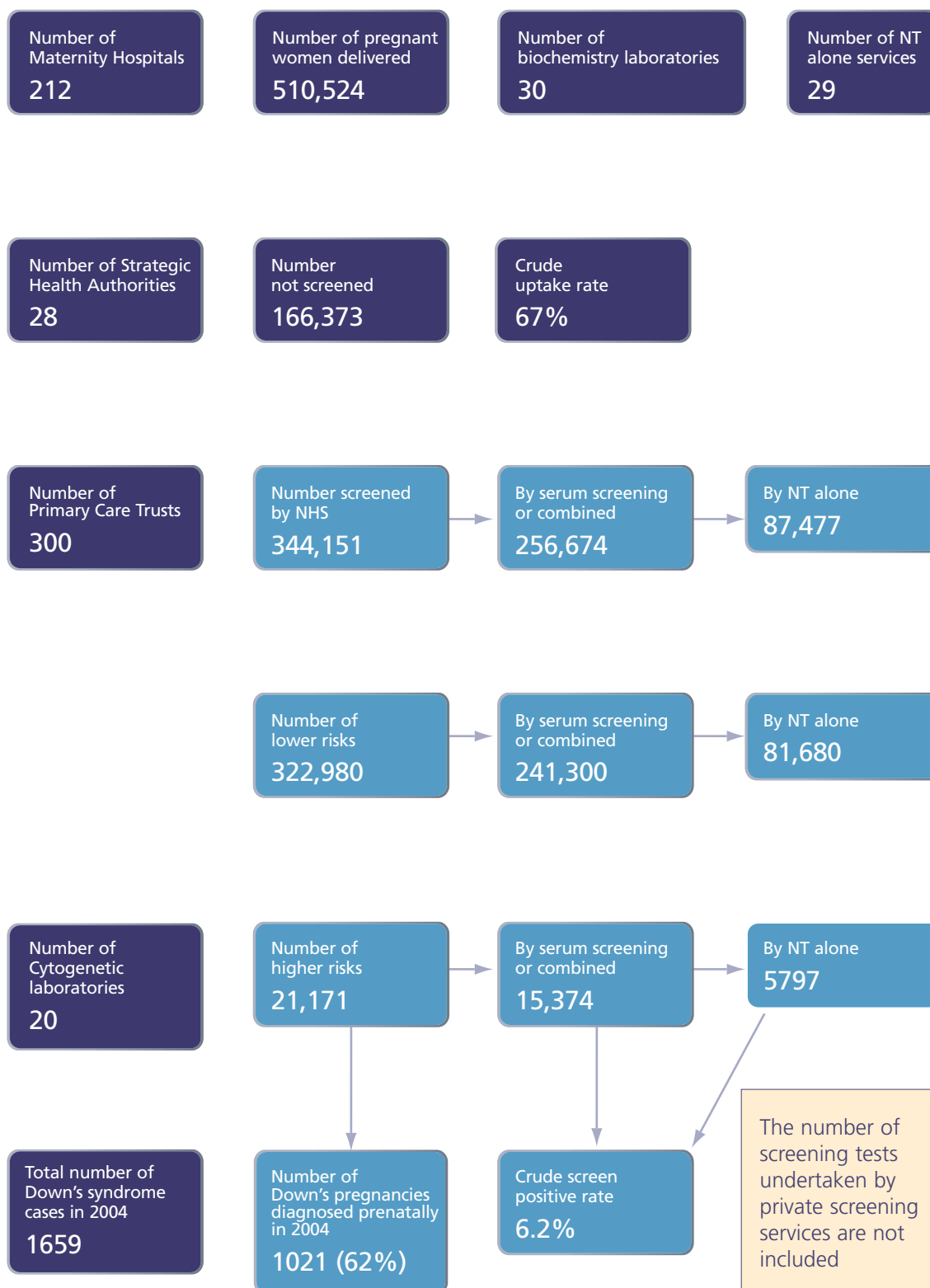
The National Down's syndrome Cytogenetic Register (NDSCR) is funded annually from the National Programme focusing particularly on the diagnosis of Down's syndrome and by what method. Specific questions have been incorporated into the questionnaire sent to local hospitals to establish the method of screening, detection and false negative rates. It has also been funded to collect information, for the first time, on the diagnosis of Trisomy 13 and 18. The data is always one year in retrospect because of the timescales of birth outcome and data retrieval. Therefore we have set out below the information available for 2004, <http://www.wolfson.qmul.ac.uk/ndscr/>

7.3 Available Data

There are 212 units undertaking maternity services, with a total of 510,524 deliveries and presently 30 biochemistry laboratories which undertake screening for Down's syndrome. This has decreased from 55 in 2002. The number of screening tests undertaken in England during 2005/2006 is 344,151. The number of screen positives generated is 21,171, which equates to a crude screen positive rate for England of 6.2%. This includes the NT alone tests which equate to 87,477. These figures can be seen in diagram 4.

1659 diagnoses of Down's syndrome were made in total for England and Wales during 2004. 1021 (62%) of those prenatally.

Diagram 4. The available statistics for the Programme for 2005 - 06



**Table 2:
The diagnosis of Down's Syndrome by outcome**

Year	Number of 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	997

Courtesy of the National Down's syndrome Cytogenetic Register

**Table 3: The number of diagnoses of
Down's Syndrome by screening method**

Year	Number of prenatal diagnoses	% of indication for Karyotyping					Median gestational age (weeks)	% of tissue sampled	
		Serum	Ultrasound <15 weeks	Serum and 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	16	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	911	33	31	0	21	13	16	37	59
2001	819	25	34	0	24	14	15	45	52
2002	898	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

Courtesy of the National Down's syndrome Cytogenetic Register

It can be clearly seen that the number of prenatal diagnoses has increased since 1989 and will most likely be increased further as full implementation is now in place. Subsequently, due to the agreement by the National Programme that screening and diagnosis based on age should not be offered, the number of women diagnosed purely using this method has dropped.

The results for 2004 showed that 25% of prenatal diagnoses were made before 13 weeks of pregnancy, 61% before 17 weeks and 14% from 17 weeks onwards.

The register has been collating information on the status of the offer of screening for those women who were given a diagnosis of Down's syndrome prenatally. This should be available more fully in the next report.

7.4 Down's Syndrome Quality Assurance Support Service (DQASS)

One other further area for collation of data is the DQASS system. The National Programme set up and funded the DQASS system at the request of laboratories, to support the quality assurance aspect. The system has been undertaken on a pilot basis from November 2004 until the present time. From 1st April 2006, at the request of the UKNSC, it will become a requirement of all laboratories to be part of this service. The aim is to be able to demonstrate adequate external quality mechanisms and enable the National Programme to make assessments and improvements on a larger scale of the efficacy of the tests after receiving data from laboratories. It will also form the basis for a national database so that continuous improvements can be made.

A total of 11 sets of data from 10 laboratories have been analysed, comprising of over 100,000 screening tests. Each set of data has been reported upon to their original laboratory and their overall detection rate and false positive rate standardised for the age of their population. The report then allows the laboratory to compare its performance against the set standard of the National Programme which is a >75% DR for a <3%FPR by 2007.

The role of the National Programme is to take forward any concerns that we have at a national level with commercial suppliers. DQASS has provided the evidence to be able to set a specification for commercial suppliers to work to that will continue to improve the results given to women. The service will be formally based at Plymouth University from 1st April 2006 and managed by Dr's. Barry Nix and Dave Wright (the statistical advisers). Further details of the DQASS service can be found on the web site <http://www.screening.nhs.uk/downs/dqass.htm> The DQASS system has been based on a similar system that operates in Toronto, Canada funded by the Ministry of Health. The 2 statistical advisers have visited Toronto to gain more knowledge about this system.

8.0 The six distinct areas of programme work

- 8.1 Inviting and informing women and their partners.
- 8.2 Professional communication.
- 8.3 National standards.
- 8.4 Programme Improvements to the testing strategies.
- 8.5 Quality assurance.
- 8.6 Education and Training.

8.1 Inviting and informing women

With full implementation of the Down's syndrome screening programme now in place all women should be offered screening prior to twenty weeks of pregnancy. This is initially undertaken by the local primary care teams which includes the midwife and the GP. The programme has provided information at the local level to assist in this area and all women must be given adequate time to consider and decide upon the offer and their choice of care pathway.

8.1.1 Patient information

The patient information group continues to review and develop the national booklet. During 2004 half a million booklets were printed and distributed throughout England. In this annual year another quarter of a million booklets have been funded and will be distributed to the service. In 2004 it was known that 74% of units use the booklet. Presently it is translated into seventeen different languages and a further two languages have been added this year at the request of the service. The national booklet is now part of Clinical Negligence Scheme for Trusts, (CNST) requirements. <http://www.nhsla.com>

New information around screening for multiple pregnancies has been developed and this will be downloaded to the service shortly. Another new area has been the collation of patient information around the diagnostic aspect of the screening process, and information and standards on this will be available during 2006.

8.1.2 DIPEX (Database Index of Patients' Experiences)

A further module of DIPEX, to be available on the web site, has been commissioned by the programme to take into account the views of women and their partners who undergo termination of the pregnancy. This gives verbal and textual accounts of the experiences of women and it is a valuable asset for those who wish to gather information in the privacy of their own home using their computer. Over 500 CDs have been distributed to the service through the conferences and newsletters. The module can be seen on the web site: www.dipex.org

8.1.3 Down's syndrome Screening Programme Web site

The website is a valuable tool to enable parents and professionals to access up to date changes and information of the National Programme. It continues to be extended. An extensive knowledge update of all published articles can be seen on the web site. It is expected that this will be reviewed annually and is available for staff. From 1st Jan 2006 to April the website had 6419 visits on its homepage. www.screening.nhs.uk/downs/home.htm

8.2 Professional Communication

8.2.1 Newsletters

The newsletter continues to be collated and printed on a quarterly basis. This is distributed to all local coordinators by the Department of Health on behalf of the Programme Centre. It has specific information about the developments in the programme. The newsletter can either be requested direct from the Programme Centre or downloaded from the web site. Feedback has been very positive on its value and we will continue to extend the contents as needed.

8.2.2 Databases

We have collated extensive contact details of all professionals at the local level with whom we need to communicate. This development has been ongoing and it allows us to distribute information to them very quickly and to receive comments on any service concerns. Regular post and email shots are sent to all professionals with specific information from the Programme Centre.

8.2.3 Conferences that have been convened by the Programme Centre

- January 2005 – The 2nd National Down's syndrome screening conference held at the Royal College of Obstetricians and Gynaecologists. 180 attendees
- June 2005 – 1st National Fetal Anomaly Ultrasound conference – including some elements of the Down's syndrome programme. 170 attendees
- July 2005 – 2 separate 2 day training courses for biochemists. One in Birmingham and one in London
- October 2005 The 2nd National Fetal Anomaly Ultrasound conference – including some elements of the Down's syndrome programme. 210 attendees
- January 2006 – Policy review forum. London, the aim of the day being to review all strategies, old and new, to enable the UKNSC to develop standards and policy post 2007. 80 invited attendees

8.3 National Standards

The working standards of the National Programme have been in place since 2004 and recently they have been extensively reviewed. Presently they are at the final consultation stage. It is expected that they will be agreed and published for late summer of 2006. The standards will incorporate new areas such as screening for multiple pregnancy, informed consent, diagnostic path ways, training and screening in private services. The standards will reflect guidance from bodies such as the Health Care Commission, <http://www.healthcarecommission.org.uk> and the Clinical Negligence Scheme for Trusts .

The Programme Centre is waiting on the report of the review by Lord Carter of Coles into laboratory services. Any recommendations from this review will be included into the new standards on reconfiguration of services. One of the major changes of the national standards will be the increase in sample throughput for the laboratories. Presently this is set at 5,000 per laboratory but will be increased to a minimum of a 10,000 samples throughput.

8.4 Programme Improvements to the Test

8.4.1 Improvements to the testing strategy

During 2004 the UKNSC commissioned work into the use of Inhibin A as a fourth marker in laboratory. A recommendation following this report from the laboratory working group was forwarded to the UKNSC in 2005. A statement issued by the UKNSC stated that Inhibin A should not be used as part of the routine laboratory work unless they have specific expertise and experience. However it is recognised that if Inhibin A is not in use then the programme will not be able to achieve the set standard of a DR>75% for a FPR<3% particularly in the second trimester.

Therefore one of the key aims for the next year is to assess other screening strategies that will be able to meet the set standard. To assess new strategies that will meet the future standards has resulted in the funding of a pilot site to look at the triage contingency screening strategy. This has been commenced in the West Midlands from 1st April 2006. The pilot should report during 2007.

There has been continued interest in 'repeated measures screening' and a request has been made to the Health Technology Assessment Programme by the Programme Centre to fund research into this particular area. <http://www.hta.nhsweb.nhs.uk/> This has been supported and proposals have been called for from the scientific community. If the completed research correlates with the original findings then the false positive rate of the screening programme should be able to be reduced to less than one per cent. This will not only have significant benefits to women going through the screening programme in that fewer will require a diagnostic test, but would also mean a reduction in the number of diagnostic procedures to be undertaken by genetics laboratories, and consequently a reduction in their workload.

8.4.2 Risk calculation software

Assessing the risk calculation software which interprets the risk given to women following a Down's syndrome screening test has been a substantial element of the work of the Programme Centre. It has been well documented by the programme that various risks can be obtained from different types of software on the same sample, and in previous reports this has been highlighted by the work of the software group. We have now reached a stage where we can set certain parameter values and have discussed these particular requirements and parameter values with commercial suppliers, and they are in the process of changing these in the software. The requirements and parameter changes will be issued in the second edition of the software specification. A copy of the specification can be obtained from the programme.

8.4.3 Machine Specification

Ultrasound machinery used for measuring Nuchal Translucency and gestational age has received very little research in the way of assessment of its ability to produce accurate measurements. As the ultrasound component is a vital part of the ability to calculate a risk for Down's syndrome the Programme Centre has funded work into assessing the capability of the various machines in the service. Dr. Tony Evans, of Leeds University, is leading on this work and it is expected that it will report in 2 years. The end product will be a specification for ultrasound machinery. This will inform the service which machines will produce a better quality image and measurement and hence improve the test.

8.4.4 Audit of ultrasound referral work

Another aspect which interfaces with the National Fetal Anomaly ultrasound programme, but will impinge on the Down's syndrome screening programme, is the available capacity in the service to implement nuchal translucency as a screening measurement. As previously stated a reason for not implementing NT has been the lack of ultrasonographers and machinery. Work will commence during the summer of 2006, to assess the possibility of reducing the number of inappropriate referrals to the ultrasound department. A number of ultrasonographers have commented that the workload could be reduced by 25%. If that is proven to be then implementing guidance on what should be referred could improve the capacity without the need to increase the workforce. The effect being that NT would be able to be introduced on a wider scale than it is now.

8.4.5 Policy review day

The Model of Best Practice (1) which forms the base policy for the National Screening Programme is due to be reviewed during 2006. It is acknowledged that recently new strategies and screening technologies have developed which require review and assessment against evidence, to see if they can be used to improve the sensitivity and specificity in the future. A policy review day was held on January 24th, 2006 in which invited experts gathered to be informed of new policies that may be available for 2008/2010. It is expected that this will form a major phase of work during 2006. The Programme Centre will be reviewing the new strategies, and evidence that supports them, to develop a future strategy which can support a lower false positive rate whilst keeping a high detection rate. It is one of the major aims of the programme to reduce the false positive rate as much as possible. As with any screening programme the emphasis is to reduce the harm as much as is possible to patients while still keeping a high performing quality test. The reduction in the amount of harm in the Down's syndrome screening programme correlates to the lowering of the diagnostic procedures that are undertaken.

8.5 Quality Assurances

8.5.1 DQASS

A major area of work during 2005/2006 has been setting up the work of DQASS (Some of this work can be seen under section 6.4). DQASS was established in a trial mode in the November of 2004 in response to feedback from laboratories, and surveys carried out by the UKNEQAS that found substantial variations in risks calculated using laboratory software. DQASS offered to audit the various stages in the risk calculation from data provided by laboratories. By the end of April 2006, eleven datasets had been processed from 10 laboratories (over 100,000 patient samples).

The trial period has identified problems both at the laboratory level and at the diagnostic company/software provider level. We have addressed this through a programme of training courses delivered by DQASS. These courses have been well received and have fostered the view that the DQASS facility can act as a catalyst in the development of more efficient and robust processes. From the National Programme's perspective DQASS completes the external quality assessment of laboratories involved in screening, augmenting the assay component being assessed by UKNEQAS. There is also emerging anecdotal evidence that other countries are becoming interested in the DQASS model for improvement, raising the possibility of greater international consistency between different risk calculation packages.

8.5.2 ROCR – Review of Central Returns (ROCR)

The Programme Centre is also working with ROCR based at the Department of Health. There will be a set number of data requirements which Trusts will be required to return on an annual basis. It is envisaged that this will commence during 2006 and a number of pieces of information such as uptake rates, detection rates and false positive rates, will be forwarded to the Programme Centre. This will begin to give a national perspective on the service.

8.5.3 Quality Assurance Group

Recently the Programme Centre has convened a new group, the Quality Assurance group. The remit is to look at specific QA issues for the programme. The group will receive aggregated reports from DQASS and any concerns raised by the Programme Director. It will also advise on ways of improving the QA areas and the process for that. It is expected that this group's remit will expand as the National Programme itself changes and encompasses all aspects of the whole programme.

8.5.4 Connecting for Health (CfH)

The National Programme continues to work with CfH to ensure that the specification set by the programme during 2004 is incorporated into their framework for the National Care Records System. Presently the Down's syndrome screening programme will be the first of the antenatal and newborn screening programme's to have its specification incorporated. It is envisaged that once this is in place then the audit function arm of the screening programme will be complete and national data will be able to be accessed by the programme producing not only an audit function but also a complete QA monitoring system of all Trusts.

8.5.5 National tariff

The National Programme has commissioned a group to develop a national tariff for Down's syndrome screening. Work has been ongoing over the past eighteen months and it is presently in the final stage of producing a base tariff. It is already acknowledged that hospitals receive a basic tariff for dating scans, mid pregnancy ultrasound scans and general biochemistry, although the biochemistry is not specific to Down's syndrome screening. Further work will clarify the base tariff for nuchal translucency measurement and specific biochemistry.

8.6 Education and Training

8.6.1 The education and training pack

In 2004, work commenced on and led to the development of, funding and distribution of the Down's syndrome screening training and education pack for professionals. 500 have been distributed to the service through the regional antenatal screening teams during 2005 and we are now awaiting the evaluation of this resource. Initial reports have been that the pack has been extremely well received. There has been very little negative comment. It will shortly be reassessed and possibly a second edition will be recommissioned for the service. The Programme Centre has received requests from different countries to use this particular training pack.

8.6.2 Biochemist training days

Four training days were setup specifically for biochemists at their request by the DQASS team. These were extremely well received and further days will be available during 2006. The aim being that the Programme Centre will produce specific information and training on the calculation and analytical aspect of Down's syndrome screening in the laboratory.

8.6.3 Nuchal Translucency training

The National Programme awarded the contract to the Fetal Medicine Foundation (FMF) in London to undertake training for nuchal translucency measurement in England. This is specifically directed at those clinicians or sonographers who will implement this particular screening test in their Trust.

To date 600 people have been trained through the NSC NT training programme and another 29 hospitals have contacted the Programme Centre since January 2006 to have a training date organised. The names of the Trusts that have undergone training will be available on our web site so that anyone can be aware of those that have received accreditation from the National Programme.

8.6.4 Resource cards

The UKNSC has commissioned and produced 33,000 resource cards which are available for all midwives to be used in their everyday work. The cards, which are of credit card size, can be carried easily by staff and show information about all screening programmes. They are designed to be an easy resource to use to help when discussing tests with women. The Programme Centre has funded part of this important resource as well as producing the specific information.

8.6.5 Gestation calculator wheel

The Programme Centre has produced its own gestation calculator wheel for midwives and sonographers to use. It has specific information about screening during the pregnancy and in particular relates to the screening time line. These have proved to be very popular with all staff. 5,000 were originally commissioned and due to their popularity more will be produced during 2006.

8.6.6 Training for sonographers

Training sonographers about some of the wider issues of communication and information giving has been undertaken by ARC (Antenatal Results and Choices) on behalf of the Programme Centre. These were very well received in 2004 so a further 9 days, one per region, have been funded for 2005/2006.

Summary

The past year has been an extremely busy one for the National Programme and its supporting staff at the Programme Centre, achieving almost a 100% implementation with the help of the Regional Antenatal Screening teams. The base quality assurance mechanisms are now in place with further development and extension of these forming the major central work for the future. There is mature development of many arms of the programme such as patient information and education which has been embedded producing a firm and permanent structure on which to build. The Programme Centre continues to work with the 6 main areas of programme function outlined in section 8, and as the Fetal Anomaly ultrasound screening programme work develops, certain aspects will dovetail and interface with the Down's syndrome screening programme to enrich it further.

Recognition is given that the audit arm is still in an embryonic state until the complete integration and connection of the national IT programme, however we will continue to access the available data collection points to produce a more comprehensive report in the future. Benchmarking against the standards will commence in 2006/2007 and will be incorporated in future reports.

Useful Websites

Department of Health and associated agencies

The Department of Health:
<http://www.doh.gov.uk/>

Health Technology Assessment:
<http://www.nchta.org/>

The National Institute for Clinical Excellence (NICE):
<http://www.nice.org.uk/>

Government website for Clinical Governance:
<http://www.cgsupport.org/>

Pathology modernisation programme:
<http://www.doh.gov.uk/pathology/modernisation/>

Medicines and Healthcare products Regulatory Agency:
<http://www.mhra.gov.uk/>

Cytogenetics

The Association of Clinical Cytogeneticists:
<http://www.cytogenetics.org.uk/information/Default.htm>

Royal Colleges

The Royal College of Pathologists:
<http://www.rcpath.org/links/>

The Royal College of Midwives:
<http://www.rcm.org.uk/>

The Royal College of General Practitioners:
<http://www.rcgp.org.uk/>

The Royal College of Obstetricians and Gynaecologists:
<http://www.rcog.org.uk/>

Parent support organisations

Antenatal Results and Choices:
<http://www.arc-uk.org/>

Contact a family:
<http://www.cafamily.org.uk/>

Support of Fetal Trisomies SOFT UK:
<http://www.soft.org.uk>

Down's Syndrome Association:
<http://www.dsa-uk.com/frameset.htm>

Quality Assurance

DQASS:
<http://www.screening.nhs.uk/downs/dqass.htm>

NEQAS:
<http://www.ukneqas.org.uk/>

Clinical Pathologists' Accreditation:
<http://www.cpa-uk.co.uk>

The Association of Clinical Biochemists:
<http://www.acb.org.uk/>

Review of Central Returns:
<http://www.ic.nhs.uk/rocr>

Information on screening

National Screening Committee:
www.nsc.nhs.uk

DIPEX. – Videos and texts of patients experiences:
www.DIPEX.Org

National Electronic Library for Health:
www.nelh.nhs.uk/screening

Information on Down's syndrome

National Down's Syndrome Cytogenetic Register:
<http://www.smd.qmul.ac.uk/solfson/ndscr/>

Down Syndrome Medical Information Group:
<http://www.dsmig.org.uk/>

Appendix 1

Membership of the Working Groups of the National Down's Syndrome Screening Programme

Down's syndrome screening Implementation Advisory Group

Prof Peter Soothill – Chair
 Dr Trish Boyd
 Dr Peter Davidson
 Dr Angus Dobbie
 Dr Henrietta Ewart
 Dr Sandy Goodburn
 Prof Jenny Hewison
 Prof Chris Price
 Dr Diana Wellesly
 Mr Paul Wood
 Dr David Worthington
 Ms Jane Fisher
 Prof Michael Model

NHS NT Training Programme Advisory Board

Dr Trish Boyd
 Dr Trish Chudleigh
 Mrs Lynne North
 Dr Colin Pollock
 Mr Paul Wood

Software Group

Dr Gordon Paterson – Chair
 Dr Barry Nix
 Dr Dave Wright
 Dr David Worthington

Laboratory Advisory Group

Prof Chris Price – Chair
 Dr Dave Worthington
 Prof Howard Cuckle
 Ms Jane Hibbert
 Dr Barry Nix
 Dr Brian Senior
 Dr Kevin Spencer
 Ms Sue Standing
 Dr David Aitken
 Mr Andy Ellis
 Dr Rick Jones
 Dr Sandy Goodburn

Patient Information Group

Prof Jenny Hewison – Chair
 Mrs Jenny Neal
 Dr Josephine Green
 Mrs Donna Kirwan
 Mrs Jane Denton
 Mrs Vicky Carne

Multiple Pregnancy Information Forum

Mrs Jane Denton - Chair
 Mrs Jenny Neal
 Dr Trish Chudleigh
 Carol Robins
 Carolyn Gleeson
 Dr Ruwan Wimalasundera
 Dr Lorin Lakasing

National Antenatal Costing Exercise

Mrs Donna Kirwan - Chair
 Dr Ellis Friedman
 Dr Brian Senior
 Mrs Rosemary Johnson
 Ms Sue Standing
 Dr Paula Grey
 Mr Martin Stanley
 Dr Guy Hayhurst
 Mr Kevin Green

Quality Assurance Group

Dr Robin Wilson – Chair
 Dr Anne Mackie
 Dr Barry Nix
 Dr Dave Wright
 Dr Brian Senior
 Dr David Worthington
 Mrs Alison Cryer
 Mr Andy Ellis
 Dr Tony Evans



