NHS Connecting for Health

Service Implementation - Do Once and Share

Clinical Genetics Action Team

Final Report

Version 2.0

Date 4.5.06

Author: Greta Westwood
I Karen Temple

SHA Lead: Jenifer Smith

Amendment History:

Version	Date	Amendment History

Forecast Changes:

Anticipated Change	When

Reviewers:

This document must be reviewed by the following. Indicate any delegation for sign off.

Name	Signature	Title / Responsibility	Date	Version
Muir Gray		Director KPS		

Approvals:

This document requires the following approvals:

Name	Signature	Title / Responsibility	Date	Version
Muir Gray		Director KPS		
DOS Programme Manager		Jayne Slater		

Distribution: Core and Action Team. (Report also for - Heads of UK Clinical Genetics Services, Specialist Commissioners, National genetics committee members)

Document Status:

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Related Documents:

These documents will provide additional information.

Ref no	Doc Reference Number	Title	Version
1	NPFIT-SHR-QMS-PRP-0015	Glossary of Terms Consolidated.doc	6

Glossary of Terms:

List any new terms created in this document. Mail the NPO Quality Manager to have these included in the master glossary above [1].

Term	Acronym	Definition
Finished Clinical Genetics Episode of Care	FCGE	The unit of clinical genetics care required to answer the patient referral question and made up of one or more Significant Contact.

Contents

Appendices

ACTION TEAM: Clinical Genetics
ACTION TEAM LEAD: Dr I Karen Temple
PROJECT MANAGER: Greta Westwood
LOCATION OF ACTION TEAM: Wessex Clinical Genetics Service, Princess Anne Hospital, Southampton
SHA LEAD: Jenifer Smith, Hampshire and Isle of Wight Strategic Health Authority
ACTION TEAM START DATE: 4th October 2006
DATE OF FINAL REPORT: 5th May 2006
1. BACKGROUND
There are 18 regional NHS genetics centres in England delivering genetic medicine. They bring together clinical geneticists, genetic counsellors and genetic laboratory scientists and provide a diagnostic and genetic counselling service. The clinical genetics arm of the service provides care for patients and relatives seeking advice about disorders with a genetic, or potentially genetic, cause. Such conditions affect all age groups from the fetus to the elderly; can involve all body systems and range from human developmental abnormalities to cancer. The genetic laboratories perform most of the DNA based genetic testing in the NHS. There are strong informal clinical genetics networks in the UK and active established professional organisations; the Clinical Genetics Society (CGS), the Association of Genetics Nurses and Counsellors (AGNC), the Association of Clinical Cytogeneticists (ACC) the Cancer Genetics Group (CGG) and the Clinical Molecular Genetics Society (CMGS) within the umbrella organisation of the British Society of Human Genetics (BSHG). There are also national genetics committees: - Joint Committee on Medical Genetics which spans the Royal Colleges of Physicians and Pathology, the Genetics Commissioning Advisory Group (GENCAG) and the UK Genetics Testing Network steering committee that all serve the NHS genetics community and have direct input into the Department of Health Genetics Unit. Although there is general professional agreement regarding the care pathway for

patients with genetic conditions, there are many local variations and no clear agreement on how to measure quality (Clinical Genetics Society, Quality Standards of a Genetics Service (www.clingensoc.org/documents 2005). It is well recognised that in order to both properly manage individual patients and to monitor activity and outcomes, the collection and availability of clinical data at every stage of the patient journey is an essential pre-requisite for the provision of a high-quality service.

At present, genetics IT systems, to monitor such information, are created locally, and are based on individual clinical genetics unit's requirements rather than a national dataset. Furthermore clinical genetics information poses a number of unique problems with regard to the ethics of sharing individual data within families and genetic knowledge that has life long implications to the family and relatives at risk. For this reason genetic databases have grown up outside of the main NHS IT systems. Following the recent work on the Output Based Specification for Genetics IT systems (www.ngrl.org.uk/manchester/publications/ IT Perspectives 2005), the time was pertinent to deliver an agreed care pathway for clinical genetics.

The lead clinician and the project manager are both based at the Wessex Genetics Service, a regional genetics centre that serves a population of approximately 3 million people. The Wessex Clinical Genetics arm of the service is based in Southampton and the Wessex Genetics laboratory, one of two National Genetics Reference Laboratories in England, is in Salisbury. Dr IK Temple, Clinical Lead is currently President of the Clinical Genetics Society and provides key links to many allied professionals in this field.

2. PROJECT OBJECTIVES

- Map out an integrated care pathway for clinical genetics services that is robust, takes into account local variability in patterns of care and serves the needs of clinicians, managers and national comparative audit

- To develop a nationally agreed clinical genetics dataset

- Secure local and national 'stakeholder' engagement, including patient involvement, to ensure both a sustainable and credible informed body of clinicians to help support the ongoing development of the NHS' IT infrastructure as it relates to genetics

- Build on work done in the genetics Output Based Specification to identify the current system specification for 'Connecting for Health' as it applies to clinical genetics

- Identify benefits for patients and clinicians
  (including the use of data in such areas as national comparative audit, and research) that are likely to result from the wider implementation of a system

in this disease area. Identify touch points between clinical genetics care pathway and those in other specialties

- Identify links to knowledge based systems like DECIPHER /Electronic Library for Health / Gene clinics /Online Mendelian Inheritance of Man OMIM/ London Dysmorphology Database

- Explore the ethical issues that separate genetics from other specialties such as life long information and sharing of individual information in families

- Identify existing work on coding in genetics linking with HSCIC and SNOMED-CT for genetics which is yet to be developed, European Coding and Classification of Rare Diseases Task Force and ICD-10 codes to identify gaps and draw together existing pieces of work.

- Link work with that of national bodies in DOH Genetics, GENCAG, BSHG, UKGTN and JCMG

SHA EXECUTIVE SUMMARY

Name: Jenifer Smith

This project team has engaged effectively with an impressive range of stakeholders in bringing this to a conclusion and delivering the project objectives. The risks to universal adoption within clinical practice are clearly set out.

The integrated care pathway will be useful to many whether or not it can be directly transferred to an electronic system, though it is hoped this will be possible.

The team has encountered some significant challenges, which they have tackled enthusiastically. As identified in the report, the coding and ethical issues will need to be taken forward if patients are to get maximum benefit from this rapidly expanding area of medical care.

4. DETAILED REPORT ON WORK WITHIN SCOPE (any outputs from this work which are not covered as specific deliverables)

National Stakeholder involvement:

- National patient stakeholder group meeting 14th December 2005
- National symposium "Ethical Issues of the Electronic Care Record of Clinical Genetics" 30th January 2006
- Heads of UK Clinical Genetics Service meeting 31st January – 1st February 2006
- National workshop of the Association of Genetic Nurses and Counsellors 21st

February 2006

- Agenda item and presentation to the Council meeting of the British Society of Human Genetics Society 9th January 2006
- Agenda item, presentation and workshop at the Council meeting of Clinical Genetics Society 8th -9th February 2006
- Agenda item and presentation to the Council meeting of the Clinical Genetics Commissioning Group (GENCAG) 23rd February 2006
- Agenda item and presentation to the Council meeting of the Joint Committee of Medical Genetics 19th January 2006
- Agenda item and presentation to the Genetics Policy Unit, Department of Health – "The future of Genepool, NeLH and Genetics 2nd March 2006
- Agenda item and presentation to the committee meeting of the Association of Genetic Nurses and Counsellors February 2006
- National patient stakeholder group meeting 7th March 2006
- Spoken presentation to the Clinical Genetics Society Conference, Glasgow, 25th March 2006.
- Advertised the posting of final drafts of the Care Pathway and Dataset for consultation on the British Society of Human Genetics website (www.bshg.org.uk) at the CGS meeting and by email to genetics health professionals through the societies
- Presentation to the Care Record Development Board 30th March 2006
- Final drafts of Care pathway and Dataset available on website of British Society of Human Genetics (www.bshg.org.uk) as consultation documents from 31st March 2006 – 14th April 2006

Evidence of national stakeholder sign off/ agreement: After all workshops and action team meetings changes to the dataset were made and circulated for comments and copies of the development of the ICP are shown in the appendix 1. All heads of Clinical Genetics Services for England received copies of the dataset and care pathway following the Heads of Service workshop and 3 comments were received and acted on. No comments were received from website consultation.

Appendices: none

Output 1: Project Plan (appendix A)

Status: Complete

Date of completion of plan: October 2005

Summary of findings:

- Organisational structure of the DOAS clinical genetics programme
- Identification of the core action team, extended action team, patient and professional stakeholders

- DOAS core action team terms of reference
- Frequency of action team meetings
- Project manager as local point of contact for all project communications
- Notes to all action team meetings following each event
- Organisation of extended action team membership and team meetings
- Organisation of regular Clinical Lead and Project Manager meetings
- Organisation of meeting for clinical leads from every English Genetics centre
- Organisation of Genetics and Ethics symposium
- Discussion of DOAS project at local and national 'stakeholder' meetings, including patient involvement, to ensure both sustainability and a credible informed body of clinicians and managers to help support the ongoing development of the NHS' IT infrastructure as it relates to genetics

Appendices: Appendix A - Project Plan

Output 2: Output 2: Clinical engagement

Status: Complete

Date of Completion: March 25th 2006

Summary of findings:

Medical Professional and Genetic Counsellor Engagement

- The core action team required time to develop the ICP and dataset prior to widespread discussion as there was not a nationally agreed dataset for clinical genetics
- Existing datasets, diagnostic coding and ICPs from all other genetics centres requested by email, but very few received.
- All Heads of Clinical Genetics Services attended a national meeting to discuss Do Once and Share Clinical Genetics ICP and dataset. Participation included; Newcastle, Liverpool, Manchester, Leeds, Sheffield, Nottingham, Exeter, Cambridge, Southampton, Oxford, London (5 centres), Birmingham and Leicester. (Scotland and Wales also attended for comparison). Karen Temple has now visited the Bristol centre that was unable to attend. Northern Ireland also was unable to attend, but represented at AGNC workshop (14/02/06).
- Workshop at CGS Council away day to discuss ICP and dataset 8th-9th February 2006
- Agenda Item on BSHG Council meeting 9th January 2006
- Agenda item on GENGAG meeting 23rd February 2006
- Agenda item on Joint Committee of Medical Genetics Royal College of Pathologists meeting19th January 2006
- Agenda item at AGNC Committee 14th February 2006

- Workshop for AGNC national representatives 21st February 2006

Spoken presentation at the Clinical Genetics Society Conference 25th March 2006, Glasgow

Appendices: Appendix B – the Consultation Process and Appendix C – a summary document used to introduce the project for committees

Output 3: Patient Engagement

Status: Completed

Date of Completion: March 2006

Summary of findings:

- The Genetic Interest Group (GIG), an umbrella group for all genetics patient groups, was a key member of the DOAS core action team
- The Cystic Fibrosis Trust were also represented in the core action team
- GIG arranged a patient stakeholder meeting to discuss the 2nd draft of the ICP and dataset with invaluable input and is constantly undated through the core team communications
- A second patient stakeholder group met in March 2006. This meeting included some of the members of the previous meeting, and some additional members.
- There was patient group representation in the Ethical Issues of the Electronic Care Record symposium

Appendices: Appendix B – the Consultation Process

Output 4: SNOMED-CT

Status: Ongoing

Date of Completion: see recommendations in report

Summary of findings:

- The benefits of national codes are discussed in the project document and compared to the position in Holland where national codes are in use
- Currently diagnostic coding for clinical genetics is different in each genetics centre. This was demonstrated by a survey of centres. Codes used by 6 centres are available but not included in this report as there is no consistency between them
- SNOMED-CT International Board member, Yves Lussier, has been contacted and SNOMED management are looking into the feasibility of including genetics within SNOMED-CT. Andrew Devereau is leading within the UK on

this topic. He is a computer scientist, member of the core action team and also represents the genetic laboratories through the Manchester National Reference Laboratory and UKGTN
- Department of Health to arrange a meeting with SNOMED-CT and Medical Genetics groups in July 2006. Date to be arranged
- Contact made with Grant Kelly (SNOMED-CT) and David Markwell (Clinical Information Consultancy) regarding coding for clinical genetics
- Prof Segolene Ayme has been contacted. She runs Orphanet – a European network for rare genetic diseases, and is creating new genetic diagnostic codes for the purposes of the network and linking them to new ICD codes. She is in touch with Andrew Devereux
- EUROCAT is a network of centres recording birth defects and they have expertise with diagnostic coding – Dr Diana Wellesley can be contacted at diana.wellesley@suht.swest.nhs.uk

Appendices: none

Output 5: Literature review

Status: Completed

Date of Completion: 31st March 2006

Summary of findings:
- Literature searching has demonstrated that little literature exists for genetic disease specific care pathways in both lay and academic publications
- There is relatively little information for referrals to clinical genetics
- A review of journal available on line within the NHS revealed that the majority of those required are not available to NHS staff

Appendices: none

Output 6: Input into "The Future of Genepool and NeLH for genetics" with the Department of Health

Status: Completed

Date of Completion: 2nd March 2006

Summary of findings:
- Presentation to the Genetics Policy Unit, Department of Health to demonstrate outcome of Do Once and Share Clinical Genetics Project
- Discussion with group about the possible future for Genepool and information learnt from Do Once and Share

Appendices: none but presentation available if required

5. UPDATE ON PROJECT CONSTRAINTS and RISKS

- The care pathway and dataset were much more challenging to produce than had been anticipated. The permutations of working practices were complex
- Genetic consultations involve thousands of different types of diseases that can be dealt with in different ways
- The family aspects of genetic medicine meant that the care pathway had to consider the relatives as well as the index case
- A care pathway and dataset have never been written in the specialty before and there was no prior experience. This meant that it took much of the project to write the care pathway and dataset prior to consultation. It has also meant that there has been less time for dissemination of the findings given the short time of the project. Much time was spent on the care pathway in committee because of the complexity and less time was available to discuss the detail of the dataset
- The dataset has not been trialled within an actual live database. It may prove too detailed for current clinical practice or may not be possible to translate into a working system
- Unnecessary time was spent on creating some code definitions that were subsequently found to already exist in other specialties, such as patient demographics. Input regarding existing NHS datasets may have helped to ensure that time was not spent trying to re invent something that was already available to the NHS. However some codes are unique to genetic medicine such as ‘family number’ and these concepts need to be adopted by the NHS for the dataset to be of use to the genetics medical community
- The word processing skills of the team have meant that the technical aspects of writing the final project has been challenging
- Time to complete the project has been the major constraint. Much of the 6 months was spent writing the ICP and dataset before it could be discussed with others. More dedicated time was needed at the end of the project to write the project report and discuss it with core team members. This has not been possible as the consultation was continuing until a few days before the project was due to finish
- The major risk is that the clinical genetics community will not translate the work into normal clinical practice because the software and computing will lag behind. Without the easy ability to enter data, staff will not be able to deal with the complexity of the dataset. While this type of coding is in use in other specialties this is not the case in Clinical Genetics where IT systems have largely grown up outside the hospital IT systems

- The DOAS clinical project team now return to normal clinical duties and the work of disseminating the work could fail. More time is required to continue to dedicate to the project and time will be needed to champion the ideas with the Heads of Clinical Genetics Service now that the report is written
- The concept of the Finished Clinical Genetics episode of Care as the unit of currency for the specialty is a new idea that has emerged out of the consultation and may be interpreted in a different way by different professionals. Recommendations are included in the final report document
- SNOMED-CT executives are not as yet sure whether the diagnostic coding systems will be suitable for Clinical Genetics Services. No national codes exist and these are essential for national datasets to be compatible
- The Care pathway may require further amendment as it is used and dedicating time to this will be difficult

6. DETAILS OF ANY CONTINGENCIES IMPLEMENTED

The report will be printed and sent to all Heads of Service and presented at all national genetics committees and national commissioning meetings for widespread use. The report is written as a stand-alone document for this purpose and includes sections that are written to improve the likelihood of the concepts of the pathway being interpreted in the same way. The need for national diagnostic codes is now recognised by the Clinical Genetics Service and is being taken forward by the Department of Health.

Recommendations for future IT requirements are summarised at the beginning of the report.

7. DELIVERABLES (progress report on all deliverables listed in the Scoping Document)

Deliverable name: Integrated Care Pathway (ICP) (see Appendix D - Do Once and Share Clinical Genetics Project Report)

Status: Completed

Date for Completion: 31st March 2006

Summary findings:

- 9 drafts of the ICP were presented monthly to the action team group concluding on March 14th. Changes were still being made to the document after this meeting. The ICP was presented to the UK Clinical Genetics

Society in March 25th 2006 and has been available for national comments from this date on the BSHG website

- The ICP were also presented to, and modified by 2 patient stakeholder groups. Patient group highlighted a) the issue of consent when tested for a genetic condition by a non-geneticist b) the timing of consent request to share family information and c) the use of an electronic version of the family history questionnaire

Stakeholder Consultation process: See report on named individuals involved in stakeholder process

Evidence of national stakeholder sign off / agreement: ICP and dataset available for national consultation on British Society of Human Genetics Society website. Prior to this it has been extensively developed following exhaustive consultation.

Issues: The major risk is that the work will not be translated into normal practice by the clinical genetics community because the software and computing will lag behind. The issues regarding differences of interpretation within the care pathway are covered within the recommendation sections after each new concept within the pathway is introduced (see report).

There has been general support for the project. Some people believe that the dataset is too complex for current IT software available in many centres.

Mitigations: No ICP had been written prior to this project for Clinical Genetics.

Appendices: Appendix D – the Do Once and Share Clinical Genetics Project Report and Appendix E – ICP development October to March 2006

Deliverable name: Ethical concerns of the electronic care record for clinical genetics

Status: Completed

Date for Completion: January 2006

Summary findings:

A national meeting to discuss the ethical issues and concerns of the electronic care record for clinical genetics took place on 30/01/06. Clinicians, medical lawyers, medical ethicists, a member of The Care Record Development Board(CRDB)/The Ethics Advisory Group, national leads in molecular genetics and cytogenetics and patients attended. Conclusions and recommendations are made in the DOAS Clinical Genetics Project Report section 3 Appendix D and as the Appendix "Ethical issues of the Electronic Care Record for Clinical Genetics" which was presented to the Care

Record Development Board.

Stakeholder Consultation process: Presentation at the national Genethics meeting November 2006, the DOAS ethics consultation January 2006 and presentation at the Care Record Development Board in March 2006.

Evidence of national stakeholder sign off / agreement: This has not been possible but the summations of the discussions are given in the report document (Appendix D under 'Ethical issues' section 3).

Issues: This topic needs further debate and it is suggested that this is taken to the Human Genetics Commission.

Mitigations: This is a very difficult subject and one that needs to be written with ethicists outside of the DOAS project. We plan to write an article for the BMJ but it is not yet written. The summary of the conclusions is included in the final report. Presentation at the Care Record Development Board initiated the discussion, which largely centres on the need for sharing of information between family members and further debate is warranted.

Appendices: Appendix D – The DOAS Clinical Genetics Project Report and Appendix F – the CRDB presentation document – 'Adapting the electronic care record to meet the needs of genetic medicine'.

Deliverable name: Agenda for SNOMED-CT

Status: on going

Date for Completion: uncertain

Summary to date: see output 4 where it has already been discussed in detail

Mitigations: The survey of centres showed that there was no national consensus and different diagnostic codes are in use throughout England. Andrew Devereux is taking this forward for the national genetics community with the Department of Health. There was not time to do this within the 6 months. The project has highlighted the need for a national consensus and the project report highlights the benefits to be gained by national agreement on coding. The DOAS report will be the basis for the discussion on what happens in the Clinical Genetics Service.

Appendix: Appendix D – the DOAS Clinical Genetics Project Report

8. ADDITIONAL INFORMATION & RECOMMENDATIONS

Future areas of work to be considered are fully addressed within the project under IT requirements in Clinical Genetics

It is assumed that software and hardware necessary for the data entry and analysis will be available. They are currently not available for Clinical Genetics Services.

Knowledge support has been identified from wide consultation

Appendices: Appendix G

9. CLINICAL LEAD COMMENTS & CONCLUSIONS

A Care pathway and national dataset for Clinical Genetics has never been attempted before and this project has achieved it. The report is the conclusion of 6 months of dedicated effort and consultation with colleagues throughout England. There has been a generally favourable response to the work and good collaborations formed within the action teams. The Care pathway records the journey through the Clinical Genetics Service from referral to discharge and has identified the dataset to accompany it. This in itself will be of use when describing the specialty to others and within Clinical Genetics, if adopted, will allow sharing of information between centres that has never previously been achieved for the purposes of research and audit.

The ethical issues related to family working have been highlighted and require continued debate. The summary and recommendations are presented within the report document. In addition to the ICP and dataset, touch points with other specialties are identified, IT knowledge requirements are included at the end of the report and benefits of national datasets are discussed. Family history is an integral part of all health care and generic tools are required for electronic pedigree drawing.

Despite the working habits within the Clinical Genetics Community being broadly comparable and networks within the profession being good, writing the Care pathway and all the complex permutations, has turned out to be far harder than predicted. I believe the current pathway must be kept under review and updated as it is used. It is a pathway that continues to develop but a starting point has now been laid down. There was not time to attempt to see whether a computer system could be designed to deliver the dataset in practice and this is a risk for the project.

The conclusions and recommendations of the project are presented in the Executive Summary of Appendix D.

10. Do Once and Share PROGRAMME COMMENTS

SIGN OFF

SHA Lead	
Name:	
Signature:	
Date:	
DOAS Programme Manager	
Name:	Jayne Slater
Signature:	
Date:	
KPS Programme Director:	
Name:	Muir Gray
Signature:	
Date:	

Appendix B - Consultation Process

	Core Action Team		
1	Dr Karen Temple	Clinical Lead/Consultant Geneticist	Wessex Clinical Genetics Service, Southampton
2	Greta Westwood	Project Manager/Genetic Counsellor	Wessex Clinical Genetics Service, Southampton
3	Jane Stephenson	Health Services Librarian	Health Services Library, University of Southampton
4	Trudi Mann	Health Services Commissioner	Somerset NHS
5	Melissa Winter	Communication Manager	Genetic Interest Group
6	Emma Wicks	Patient Advisor	Cystic Fibrosis Trust
7	Veronica Price	General Practitioner	Eastney Health Centre, Portsmouth
8	Andrew Devereau	Health Services Informatics	National Genetics Reference Lab, Manchester
9	Linda Howard	Programme Manager	Connecting for Health
	Extended Action Team		
10	Dr Jonathon Berg	Consultant Geneticist	University of Aberdeen
11	Dr Phil Zack	Consultant Geneticist	Guys and St Thomas' Hospital, NHS Trust, London
12	Dr Helen Firth	Consultant Geneticist	East Anglian Regional Genetics Service, Cambridge
13	Dr Carol Chu	Consultant Geneticist	Yorkshire Regional Genetics Service, Leeds
14	Jo Haydon	Genetic Counsellor	West Midlands Regional Genetics Service, Birmingham
15	Jan Moore	Genetic Counsellor	West Midlands Regional Genetics Service, Birmingham
16	Jenifer Smith	Strategic Health Authority Lead	Hampshire and IOW SHA
17	Lyn Fox	Manager	Leicestershire Genetics Centre, Leicester
18	Alison Hill	Genetics Policy Unit	Department of Health, London
	Patient Group meeting 14th December 2005		
19	Emma Wicks	Expert Patient Advisor	Cystic Fibrosis Trust
20	Melissa Winter	Communication Manager	Genetic Interest Group
21	Lyn Zwink	Chair	Fragile X Society
22	Mrs Howie	Representative Member	Cerebellar Ataxia Group
23	Catherine Howie	Representative Member	Cerebellar Ataxia Group
24	Jo Gray	Committee member and Trustee-	AMEND
	Genethics Symposium 30th January 2006		
25	Anneke Lucassen	Consultant Clinical Geneticist	Wessex Clinical Genetics Service, Southampton
27	Ian Ellis	Consultant Clinical Geneticist	Cheshire and Merseyside Genetics Service, Liverpool
28	Tara Clancy	Genetic Counsellor/Lecturer	Regional Genetics Service, Manchester

29	Alison Hall	Research Associate	Cambridge Genetics Knowledge Park
30	Mark Bale	Genetics Policy Unit	Department of Health, London
31	Ron Zimmern	Director of Public Health Genetics Unit	Cambridge Genetics Knowledge Park
32	Robert Wheeler	Consultant Paediatric Surgeon	Southampton General Hospital
33	Michael Parker	Professor of Medical Ethics	The Ethox Centre, Oxford
34	Fiona Douglas	Honorary Clinical Lecturer	Northern Genetics Service, Newcastle
35	Angus Clarke	Consultant Clinical Geneticist	Institute of Medical Genetics, Cardiff
36	Joan Higgins	Chair of Ethics Advisory Group	Care Record Development Board, CfH
37	Melissa Winter	Communications Manager	Genetic Interest Group
38	Fiona Lalloo	Clinical Geneticist	Regional Genetics Service, Manchester
39	David Robinson	Molecular Geneticist	Wessex Genetics Reference Lab, Salisbury
40	Teresa Davies	Clinical Cytogeneticist	South Western Regional Genetics Service, Bristol
41	Christina Honeywell	Cardiovascular Genetic Counsellor	Oxford Regional Genetics Service
42	Carol Chu	Clinical Geneticist	Yorkshire Regional Genetics Service, Leeds
43	Rick Jones	Health Informatics, NPFiT	Leeds
44	Tessa Shellens	Lawyer	Cardiff
45	Delyth Alldrick	Chair Clinical Ethics Committee	Cardiff
46	Jane Kaye	Research Fellow	Ethox Centre, Oxford
47	Sean Kirwan	Policy Development Manager	Digital Information Policy Team, CfH
	Heads of Clinical Service 31st January 2006- 1st February 2006		
48	Alan Fryer	Consultant Clinical Geneticist	Cheshire and Merseyside Genetics Service, Liverpool
49	Anneke Lucassen	Consultant Clinical Geneticist	Wessex Clinical Genetics Service, Southampton
50	Mandy Collins	Consultant Clinical Geneticist	Wessex Clinical Genetics Service, Southampton
51	Carole Gardiner	Consultant Clinical Geneticist	Nottingham Genetics Service
52	Ros Eles	Consultant Clinical Geneticist	Royal Marsden Hospital NHS Trust, London
53	Carole Brewer	Consultant Clinical Geneticist	Peninsula Clinical Genetics Service, Exeter
54	Annie Proctor	Consultant Clinical Geneticist	All Wales Medical Genetics Service
55	Frances Elmslie	Consultant Clinical Geneticist	South West Thames Regional Genetics Service, St Georges Hospital
56	Bronwyn Kerr	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
57	Helen Stewart	Consultant Clinical Geneticist	Oxford Regional Genetics Service
58	Angela Barnicoat	Consultant Clinical Geneticist	North East Thames Regional Genetics Service, Great Ormond Street, London
59	Carol Chu	Consultant Clinical Geneticist	Yorkshire Regional Genetics Service, Leeds
60	Jackie Cook	Consultant Clinical Geneticist	North Trent Genetics Centre, Sheffield
61	Michael Wright	Consultant Clinical Geneticist	Northern Genetics Service, Newcastle
62	Shela Mohammed	Consultant Clinical Geneticist	Guys and St Thomas' NHS Trust, Department of Clinical Genetics
63	Trevor Cole	Consultant Clinical Geneticist	West Midlands Regional Genetics Service, Birmingham
64	Joan Patterson	Consultant Clinical Geneticist	East Anglian Regional Genetics Service, Cambridge
65	Margaret Barrow	Consultant Clinical Geneticist	Leicestershire Genetics Centre, Leicester

66	Sue Holder	Consultant Clinical Geneticist	North West Thames Regional Genetics Service
	Clinical Genetics Society council February 8th – 9th 2006		
67	Peter Farndon	Consultant Clinical Geneticist	West Midlands Regional Genetics Service, Birmingham
68	Helen Kingston	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
69	Chris Bennett	Consultant Clinical Geneticist	Yorkshire Regional Genetics Service, Leeds
70	Paul Brennan	Consultant Clinical Geneticist	Northern Genetics Service, Newcastle
71	John Burn	Consultant Clinical Geneticist	Northern Genetics Service, Newcastle
72	Jill Clayton Smith	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
73	John Dean	Consultant Clinical Geneticist	North of Scotland Regional Genetics Service, Aberdeen
74	Dian Donnai	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
75	Gareth Evans	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
76	Bronwyn Kerr	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
77	Fiona Lalloo	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
78	Peter Lunt	Consultant Clinical Geneticist	South Western Regional Genetics Service, Bristol
79	Sahar Mansour	Consultant Clinical Geneticist	South West Thames Regional Genetics Centre, St Georges Hospital, London
80	Adam Shaw	Consultant Clinical Geneticist	South West Thames Regional Genetics Centre, St Georges Hospital, London
81	Audrey Smith	Consultant Clinical Geneticist	Regional Genetics Service, Manchester
82	Richard Trembath	Consultant Clinical Geneticist	Guys and St Thomas' NHS Trust, Department of Clinical Genetics
83	Peter Turnpenny	Consultant Clinical Geneticist	Peninsula
84	Oliver Quarrell	Consultant Clinical Geneticist	Peninsula Clinical Genetics Service, Exeter
85	Michael Wright	Consultant Clinical Geneticist	North Trent Genetics Centre, Sheffield
	AGNC Workshop 21st February 2006		
86	Georgie Hall	Genetic Counsellor	Regional Genetics Service, Manchester
87	Marion Macallistair	Genetic Counsellor	Regional Genetics Service, Manchester
88	Nicola Crawford	Genetic Counsellor	Genetics Service, Sheffield
89	Aoife Bradlley	Genetic Counsellor	Northern Ireland Regional Genetics Service, Belfast
90	Lyn Magerison	Genetic Counsellor	North West Thames Regional Genetics Service,
91	Carole Cummings	Genetic Counsellor	North West Thames Regional Genetics Service
92	Patricia Finnemore	Genetic Counsellor	Wessex Clinical Genetics Service, Southampton
93	Sally Watts	Genetic Counsellor	Guys and St Thomas' NHS Trust, Department of Clinical Genetics
94	Carolyn Redman	NF Specialist Advisor/	Wessex Clinical Genetics Service, Southampton
95	Eva Mazeem	Genetic Counsellor	Oxford Regional Genetics Service
96	Sarah Durrell	Genetic Counsellor	Oxford Regional Genetics Service

97	Kath Smith	Genetic Counsellor	Taunton Genetics Service
98	Cathy Watt	Genetic Counsellor	West of Scotland Regional Genetics Service, Glasgow
99	Liz France	Genetic Counsellor	All Wales Medical Genetics Service
100	Linda Howard	Programme Manager	Connecting for Health
	Patient group meeting 7th March 2006		
101	Emma Wicks	Expert Patient Advisor	Cystic Fibrosis Trust
102	Melissa Winter	Communication Manager	Genetic Interest Group
103	Lyn Zwink	Chair	Fragile X Society
104	Beverley Searle	Chief Executive Officer	UNIQUE- Rare Chromosome Disorder Support Group
105	Jo Gray	Committee Member and Trustee	AMEND
106		Officer	SADS UK
107	Elaine Miller	Officer	U.K. Thalassaemia Society

Appendix C

Do Once and Share - Connecting for Health
Clinical Genetics

Introduction

The DOAS (Do Once and Share) programme is part of the NHS Connecting for Health (NPfIT) programme. The NPfIT programme will bring modern computer systems into the NHS to improve patient care and services. Over the next ten years, the National Programme for IT will connect over 30,000 GPs in England to almost 300 hospitals and give patients access to their personal health and care information, transforming the way the NHS works.

DOAS aims to enhance and inform the development of these new NHS IT systems to meet the up-to-date requirements of health services for patients. The overall objectives of the DOAS programme are to;

- develop a common approach to common conditions
- reduce duplication
- create national consistency
- reduce the waste of professional and patient time

There are 44 DOAS projects at the present time, covering the major causes of human disease in the country.

Genetic disorders are an important cause of ill health and the practical and ethical issues of organising electronic records for families have been identified as a crucial area, overlapping many other NHS services. A DOAS project for clinical genetics has been established, with the aim of writing a care pathway for patients referred to the clinical genetic service, highlighting the areas where patient care can be enhanced by IT developments.

Clinical Standards for a Genetics Unit have been recently published (www.clingensoc.org) and although there is general professional agreement regarding the care for patients with genetic conditions there are many local variations and no clear agreement on how to measure quality or process. It is well recognised that in order to both properly manage individual patients and to monitor activity and outcomes, the collection and availability of clinical data at every stage of the patient journey is an essential pre-requisite for the provision of a high-quality service. However genetics IT systems to monitor such information tend to be created locally and be based on individual unit requirements rather than a national data set. This makes comparison of data difficult. Furthermore Clinical Genetic information poses a number of unique problems with regard to the ethics of sharing individual data within families and genetic knowledge that has life long implications to the family and relatives at risk and for this reason genetic databases have grown up outside of the main NHS IT systems. Following the recent work

on the Output Based Specification for Genetics IT systems (IT Perspectives, September 2005) the time is pertinent to deliver an agreed dataset based on the patient journey through clinical genetics.

Scope of the project

- Map out an integrated care pathway with data points at each stage that is robust, takes into account local variability in patterns of care and serves the needs of health professionals, managers and national comparative audit.
- Secure local and national 'stakeholder' engagement to ensure both sustainability and a credible, informed body of clinicians to help support the ongoing development of the NHS's IT infrastructure as it relates to genetics
- Build on work done in the genetics Output Based Specification to identify the current system specification for 'Connecting for Health' as it applies to clinical genetics,
- Identify benefits for patients, clinicians and managers (including the use of data in such areas as national comparative audit, and research) that are likely to result from the wider implementation of a system in this disease area.
- Identify touch points between clinical genetics care pathway and those in other specialities
- Identify links to knowledge based systems like DECIPHER /Electronic Library for Health / Gene clinics /Online Mendelian Inheritance of Man OMIM/ London Dysmorphology Database
- Explore the ethics that separate genetics from other specialities such as life long information and sharing of individual information in families
- Identify gaps in current coding and provide an agreed agenda for SNOMED
- Link work with existing bodies in DOH Genetics, GENCAG, BSHG, UKGTN and JCMG

Out of scope

- Engage with other chronic disease programs
- Development of guidelines for individual genetic conditions
- Solving ethical dilemmas identified in clinical genetics
- Creation of diagnostic codes
- Road-testing or wide-spread implementation

The project dates - October 2005 - May 2006
Clinical lead - IK Temple, Project Manager – G Westwood.