Welcome to the DECIPHER database
An introduction for Families

Why was the DECIPHER database developed?
DECIPHER was developed to try to use the benefits of the human genome project to increase knowledge about rare chromosome disorders caused by subtle changes in the chromosomes. These changes are often too small to see using a microscope, but special laboratory techniques eg. microarrays may show that there is too little chromosomal material (microdeletion) or too much (microduplication).

What is the purpose of the DECIPHER database?

- Increasing medical and scientific knowledge about chromosomal microdeletions / microduplications
- Improving medical care and genetic advice for individuals/families with subtle chromosomal imbalance
- Facilitating research into the study of genes which affect human development and health

Who is DECIPHER for?
Decipher was developed as a tool for clinical geneticists (doctors who specialise in genetic conditions such as chromosome disorders), cytogeneticists (scientists who analyse chromosomes) and molecular biologists (scientists who study the way genes behave and how they affect health and development). In order to promote progress in medical and scientific understanding of these conditions, the fully anonymous data held in DECIPHER-Ensembl view is made available via the Ensembl genome browser on the worldwide web.

What data will be collected about me/my child?

- Information about your chromosome patterns, as tested by microarray analysis*****, or other molecular techniques, including any ways in which your pattern differs from the average.
- Clinical information which your geneticist thinks could be relevant to these chromosome variations. These may for example be physical anomalies (which are present from birth) or developmental problems/learning difficulties.
- You can decide whether you wish a photograph to be included in the entry. If permission for a photograph is given, clinicians/clinical cytogeneticists registered with DECIPHER would be able to see the photograph; it would not be visible to anyone else. It is not necessary for a photograph to be included; this is entirely optional.

With your consent, this information will be entered in DECIPHER together with details about the precise nature and location of the chromosome change. Every care has been taken in the design of DECIPHER to protect the privacy of patients. Due to the rarity of many chromosomal disorders, DECIPHER is an international project. Data Protection legislation is uniform throughout the European Union, but may vary in non-EU countries.

The DECIPHER array project*
For many patients, the microarray analysis which provides information about your/your child's chromosome pattern will be run as a clinical investigation or as part of a separate ethically approved research project. However, because of the difficulty accessing these highly specialised and expensive investigations, a number of array studies will be run as part of the DECIPHER project. If run as part of the DECIPHER array study, DNA from you/your child will be analysed by scientists at the Wellcome Trust Sanger Institute and/or by your local laboratory. Parental samples may be required in order to interpret the results of array-CGH testing. Comparison of results from parents and their child, in this level of detail, has the potential to reveal non-paternity or non-maternity. If you intend to answer 'Yes' to question 1b of the DECIPHER Consent form (stating that you would like the results of the array analysis to be shared with you), **it is essential that you have fully considered this possibility with your/your child's Clinical Geneticist before entering the study.**

How will this be displayed?
The information will be in a report (see over) and also on a computer diagram, as a coloured bar over the section of the chromosome where material has been lost or gained. The medical features will be displayed alongside this bar, so that doctors and clinical scientists can compare records to build up knowledge and understanding about specific chromosome changes and their consequences.

Why is a database like this needed?
Subtle chromosome changes can occur anywhere throughout the genome. Individual changes are therefore very rare. Bringing information together, so that doctors and scientists can share information, accelerates the path to knowledge and understanding about rare conditions.

Future contact

If your clinician is contacted by another clinical geneticist who through DECIPHER has identified other individuals with the same/similar chromosomal change and the same/similar clinical features, you may be contacted and asked whether you wish to give permission for further details to be exchanged with a view to furthering understanding of this chromosomal change. You may also be asked whether you wish anonymous details to be included in a publication in the medical/scientific literature.

Can I withdraw from DECIPHER?

If in the future you wish to withdraw you/your child's record from DECIPHER, for any reason, this can be done quite simply by making a request to your geneticist who can arrange for you/your child's record to be deleted from the database. Parents/guardians who have given consent for their child's record to be entered in DECIPHER should ensure that when the child reaches the age of 16 yrs he/she is made aware of his/her DECIPHER entry so that he/she can make his/her own choice whether to continue or withdraw the entry.

Where can I find out more?

Further information about the DECIPHER database is available on the internet at: http://decipher.sanger.ac.uk

Other questions

In this leaflet we have tried to answer any questions you may have about the DECIPHER database. If you have more questions, please ask your geneticist for further explanation. If you still have queries after speaking with your geneticist, please contact Dr Helen Firth on Tel: 01223 – 216446.

Sample report

What do I do next?

If you are willing for information about you/your child to be entered in DECIPHER **please complete and sign the DECIPHER consent form** and return it to your geneticist.

Great care has been taken in the design of DECIPHER to protect patient privacy. Clinical contributors are bound by professional codes of conduct, however the organisers can accept no legal liability for the use or misuse of information held in the database.

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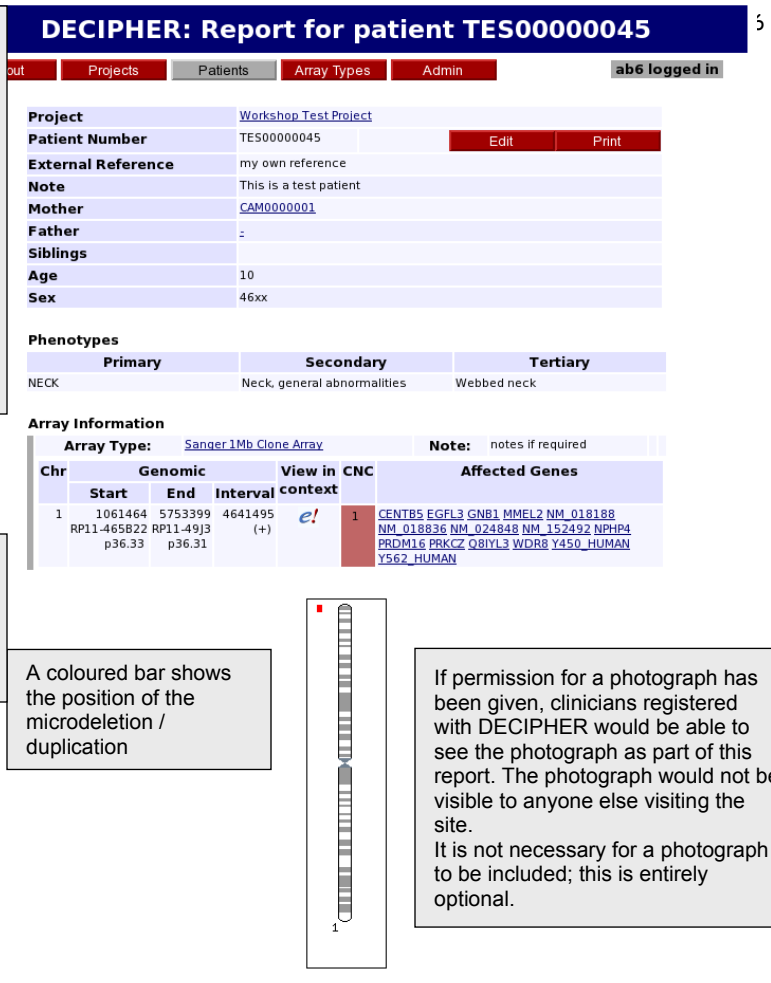
In order to protect anonymity, neither the name nor date of birth is displayed. If other members of the family have been tested and also carry a chromosome change, a link to their records is given here. Fields relating to family members are only viewable by registered clinicians who are logged into DECIPHER

Medical features (phenotypes) are listed here

Details about the microdeletion / duplication and its precise location on the chromosome are given here.

A coloured bar shows the position of the microdeletion / duplication

If permission for a photograph has been given, clinicians registered with DECIPHER would be able to see the photograph as part of this report. The photograph would not be visible to anyone else visiting the site.
It is not necessary for a photograph to be included; this is entirely optional.



DECIPHER: Report for patient TES00000045

out | Projects | Patients | Array Types | Admin | ab6 logged in

Project	Workshop Test Project
Patient Number	TES00000045 [Edit] [Print]
External Reference	my own reference
Note	This is a test patient
Mother	CAM0000001
Father	-
Siblings	
Age	10
Sex	46xx

Phenotypes

Primary	Secondary	Tertiary
NECK	Neck, general abnormalities	Webbed neck

Array Information

Array Type: Sanger 1Mb Clone Array **Note:** notes if required

Chr	Genomic Start	Genomic End	Interval	View in context	CNC	Affected Genes
1	1061464 RP11-465B22 p36.33	5753399 RP11-49J3 p36.31	4641495 (+)	e!	1	CENTB5 EGFL3 GNB1 MMEL2 NM_018188 NM_018836 NM_024848 NM_152492 NPHP4 PRDM16 PRKCZ Q8IYL3 WDR8 Y450_HUMAN Y562_HUMAN

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DECIPHER

DatabasE of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources
A collaborative venture to assign phenotype to locus on the human genome map

The DECIPHER database of submicroscopic chromosomal imbalance collects clinical information about chromosomal microdeletions/duplications and inversions and displays this information on the human genome map with the aims of:

- Increasing medical and scientific knowledge about chromosomal microdeletions/duplications and inversions
- Improving medical care and genetic advice for individuals/families with submicroscopic chromosomal imbalance
- Facilitating research into the study of genes which affect human development and health

Further information about the DECIPHER database of chromosomal microdeletion/duplications/inversions is available on the internet at: http://decipher.sanger.ac.uk

Consent Form

Please print and keep the signed copy of this form in the patient record (do not send to DECIPHER)

I give permission on behalf of myself/my child

…….…… (insert name of subject)

born……..……(insert date of birth of subject)

to Dr/Prof…….…… (insert name of clinician)

of the …….……

……..…(insert Departmental address of clinician)

to submit:

- Anonymous clinical data regarding me / my child to The DECIPHER Database — Yes/No (please circle your choice)
- Photographs of me / my child to The DECIPHER Database (optional) — Yes/No (please circle your choice)
- I have read the explanatory leaflet 'Welcome to the DECIPHER database - Information for families' — Yes/No

I understand that:

- My name / my child's name will not be published.
- The material will be placed on DECIPHER's world-wide website at http://decipher.sanger.ac.uk . The website will be seen and used by doctors (especially clinical geneticists) and scientists (especially cytogeneticists and molecular biologists). Fully anonymised summary data held in DECIPHER will be viewable via the publicly accessible Ensembl genome browser.
- Any photographs will be password protected and only available to clinicians/clinical cytogeneticists who are registered members of the DECIPHER Consortium

Signature of patient/parent/guardian…….....

Please state your relationship to the subject eg. self / mother / father /guardian …….

Date of signature…….

Signature of clinician seeking consent…….

Date of clinician's signature…….....

Thank you for taking the time to read and complete this form. Please return it to your geneticist.

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