Sample report

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

Phenotype terms are selected interactively from the London Neurogenetics and Dysmorphology database

Details about the microdeletion / duplication, its chromosomal location and approximate size in Mb are given here. Clicking on the ***e!*** button allows you to view your patient's deletion/duplication in its genomic context on the Ensembl browser and to see immediately whether any patients with a similar imbalance are known to DECIPHER, and if so, what is their phenotype.

DECIPHER: Report for patient TES00000045

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

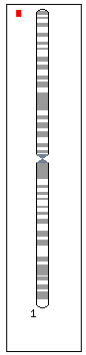
Primary	Secondary	Tertiary
NECK	Neck, general abnormalities	Webbed neck

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

A coloured bar shows the position of the microdeletion / duplication. Red denotes a deletion and green a duplication.



Genes involved in the microdeletion / duplication are shown here, together with a list of OMIM genes and links through to OMIM and Ensembl for further details about genes of interest

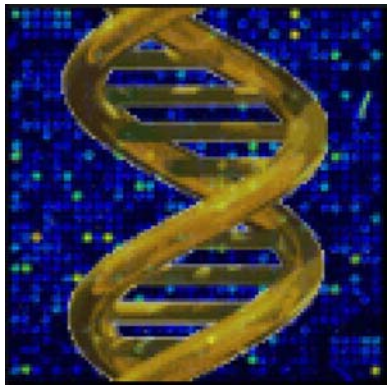
If permission for a photograph has been given, clinicians registered with DECIPHER would be able to see the photograph as part of this report. The photograph would not be visible to anyone else visiting the site.
It is not necessary for a photograph to be included; this is entirely optional.

Where can I find out more?

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

HF v.3 10/04

DECIPHER



A molecular cytogenetic database for clinicians and researchers linking genomic microarray data with phenotype using the Ensembl genome browser

http://decipher.sanger.ac.uk

DECIPHER is an acronym for **D**atabase of **C**hromosome **I**mbalance and **P**henotype in **H**umans using **E**nsembl **R**esources. The New Shorter Oxford English Dictionary (OUP 1993) defines decipher as 'to give the key to, to discover the meaning of (something obscure and perplexing)'.

Why was the DECIPHER database developed?

- Subtle chromosome changes can occur anywhere in the genome, but particular changes are very rare. Bringing information together so that it can be shared by clinicians and scientists will accelerate progress towards understanding of rare conditions and of gene function.

What does the DECIPHER database do?

- DECIPHER uses the sequence data of the human genome project to grasp the opportunity afforded by new high throughput molecular techniques particularly genomic microarray analysis to increase knowledge about submicroscopic chromosomal imbalance.
- DECIPHER allows differentiation between pathogenic and polymorphic copy number changes identified on genomic array analysis.
- By associating accurate genomic location with phenotype, DECIPHER is a powerful tool in the effort to find the function of genes of unknown function, particularly those which affect human development.

Who is DECIPHER for?

- DECIPHER is a tool for clinical geneticists, cytogeneticists and molecular biologists in the genomic microarray era. Fully anonymised summary data held in DECIPHER will be viewable via the publicly accessible Ensembl genome browser.

What are the benefits of DECIPHER to clinical geneticists?

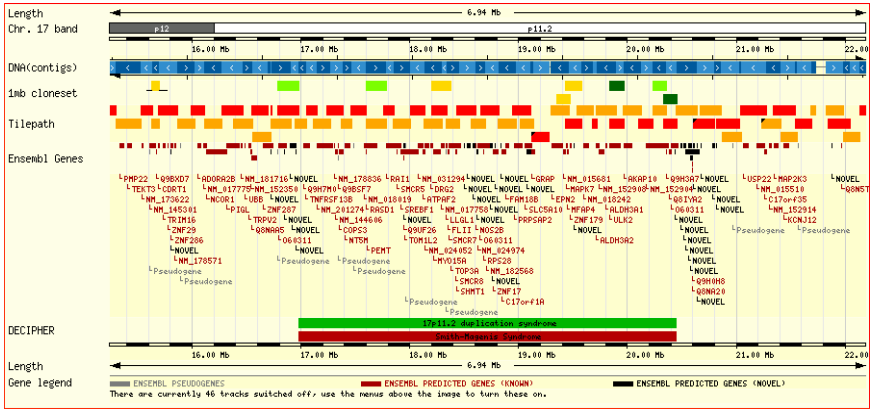
- DECIPHER shows you immediately whether a similar chromosomal imbalance has been reported previously and tells you the phenotype of the individuals concerned. This is of enormous value in determining the potential significance of copy number changes identified on an array.
- DECIPHER lists all of the known and predicted genes that are implicated in an altered region, including the OMIM genes. Genes of established high clinical significance, eg. known tumour suppressor genes, can thus be identified immediately.
- As well as showing the existence of patients with the same/similar microdeletions/duplications, DECIPHER facilitates contact with responsible clinicians in contributing centres, thereby accelerating the recognition and publication of novel syndromes
- DECIPHER provides information regarding known microdeletion/duplication syndromes and polymorphisms together with links to the relevant literature and support groups

What are the benefits of DECIPHER to cytogeneticists?

- Ability see immediately the chromosomal location of a deletion/duplication identified by genomic array analysis
- Ability to view the microdeletion/duplication in the Ensembl genome browser alongside the tiling path cloneset to facilitate selection of clones for FISH verification of the rearrangement
- Ability to generate a written report from the array data or FISH data with a karyotypic ideogram to facilitate communication with clinicians and patients

What are the benefits of DECIPHER to researchers?

- DECIPHER will allow progress in understanding the genetic basis of the phenotypes associated with copy number changes, opening opportunities for



gene identification and for refining molecular dysmorphology.

Policy for publication of data held in DECIPHER

- Any publication which refers to data contained in the DECIPHER database should acknowledge the Decipher Consortium.
- For any publication about a single chromosomal locus, the instigator should contact the main contact for the participating centre of any patient whose summary data they wish to include in their report and offer appropriate agreed recognition of their contribution, which may include co-authorship if the magnitude of the contribution warrants it to at least one representative from the participating centre (preferably the member who submitted the patient data). Where appropriate the local clinician will seek further consent from the patient eg. to exchange more detailed clinical information and/or consent for publication.