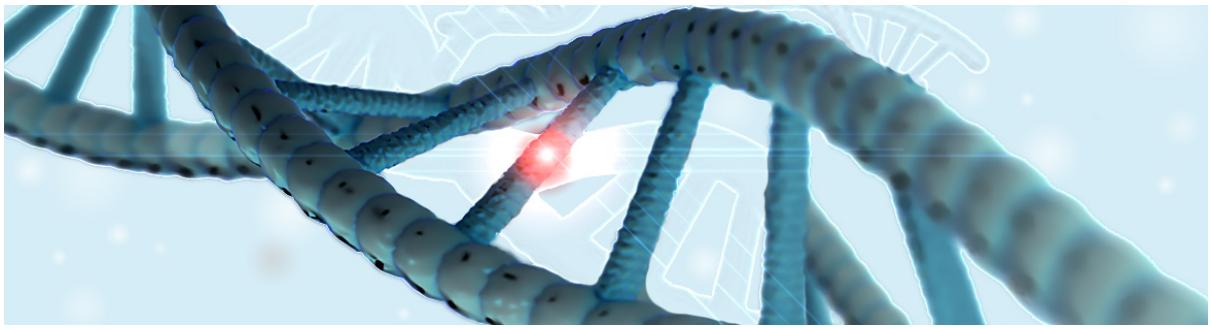
# Welcome to the DECIPHER Database



# An introduction for Families

**Why was the DECIPHER database developed?**
DECIPHER was developed to use the benefits of the human genome project to increase knowledge about rare genetic disorders. These changes are often too small to see using a microscope, but special laboratory techniques e.g. microarrays or sequencing may show that there is too little chromosomal material (deletion) or too much (duplication) or an alteration in the genetic code (sequence variant).

**What is the purpose of the DECIPHER database?**

- Increasing medical and scientific knowledge about rare genomic variants
- Improving medical care and genetic advice for individuals/families with rare genetic disorders
- Facilitating research into the study of genes that affect human development and health to improve diagnosis, management and therapy.

**Who is DECIPHER for?**
Decipher was developed as a tool for clinical geneticists (doctors who specialise in genetic conditions such as chromosome disorders), genetic scientists (scientists who analyse chromosomes & DNA) 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 and to develop new therapies, the linked anonymous data held in DECIPHER-Ensembl view is made available via the Ensembl genome browser and other genome browsers on the worldwide web. This may include the sharing of the data with other approved research institutions, universities and companies worldwide to improve care and develop and evaluate new therapies for patients with rare diseases.

**What data will be collected about me/my child**

- Information about your chromosome pattern or DNA sequence, as tested by microarray analysis, or sequencing techniques, including ways in which your pattern differs from the reference (usual) sequence.
- Clinical information which your geneticist thinks could be relevant to these rare genomic variants. These may for example be physical anomalies (which are present from birth) or developmental problems/learning difficulties.
- You can decide whether you wish an image to be included in the entry. Images will be password protected and not shared openly but can be used to generate a composite image (an average image formed from 10 or more individuals) from which any individual cannot be identified. This allows individual patients to contribute data to a composite image that can be shared safely to improve clinical diagnosis.

With your consent, this information will be shared via DECIPHER together with details about the precise nature and location of the genomic change. Every care has been taken in the design of DECIPHER to protect the privacy of patients. Due to the rarity of many genomic variants, DECIPHER is an international project.

**How will this be displayed?**
The information will be in a report (see over) and also on a computer diagram, as a coloured line over the section of the chromosome where genetic material has been lost or gained or altered. 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 genomic changes and their consequences.

**Why is a database like this needed?**
Rare genomic variants can occur anywhere throughout the genome. Individual changes are often very rare or novel. Bringing information together, so that doctors and scientists can share information, accelerates the path to knowledge and understanding about rare conditions. DECIPHER data will be shared with other genomic databases dedicated to improved interpretation of genomic variation.

**Future contact**
If your clinician is contacted by another clinician /scientist who through DECIPHER has identified other individuals with the same/similar genomic variant 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 genomic variant. You may also be asked whether you wish anonymous details to be included in a publication in the medical/scientific literature.

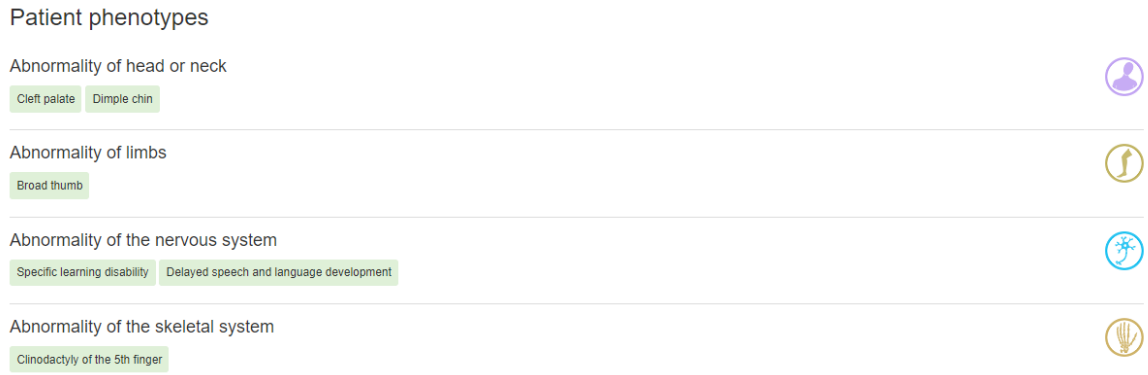
**Can I withdraw from DECIPHER?**
If in the future you wish to withdraw your/your child's record from DECIPHER, for any reason, this can be done quite simply by making a request to your geneticist who can arrange for you/your child's record to be deleted from the database. Parents/guardians who have 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 DECIPHER is available at: https://deciphergenomics.org

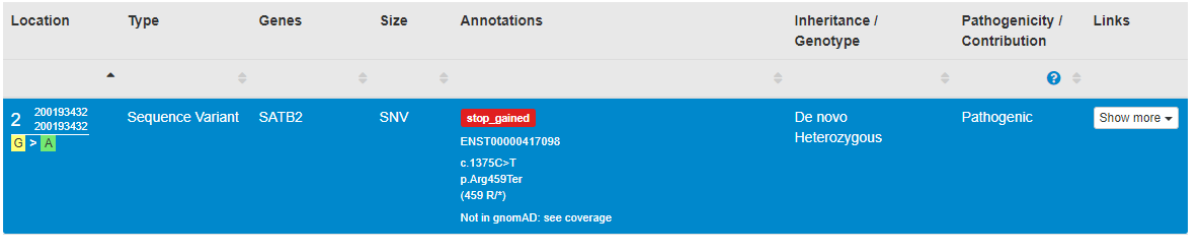
**Other questions**
In this leaflet we have tried to answer any questions you may have about the DECIPHER database. If you have more questions, please ask your geneticist for further explanation. If you still have queries after speaking with your geneticist, please contact Dr Helen Firth on Tel: 01223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

**Sample report**
DECIPHER No. 123456
In order to protect anonymity, neither the name nor date of birth is recorded in DECIPHER.

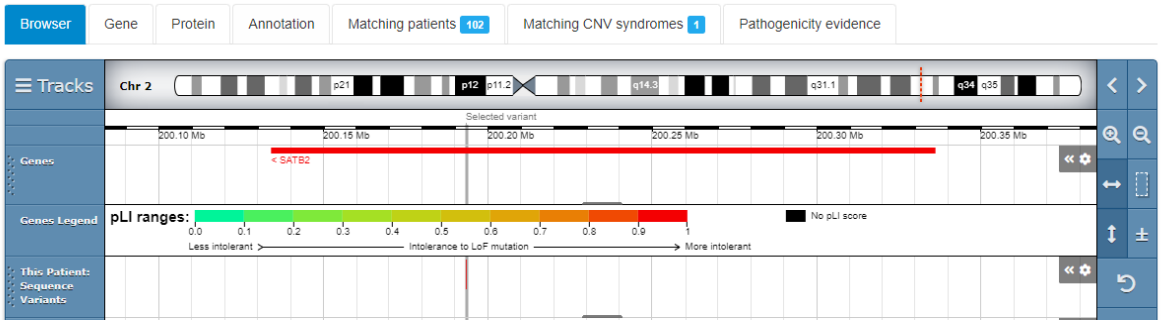
Medical features (phenotypes) are listed here:




Details about the genomic variant and its precise location on the chromosome and inheritance (if known) are given here:




A coloured bar shows the position of the genomic variant as shown here:




Details about the genes involved in the genomic variant can be accessed to guide clinical management.
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.

**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 research ethics 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.