

SEPT
2012

DECIPHERING DEVELOPMENTAL DISORDERS FIRST ANNUAL FAMILY NEWSLETTER



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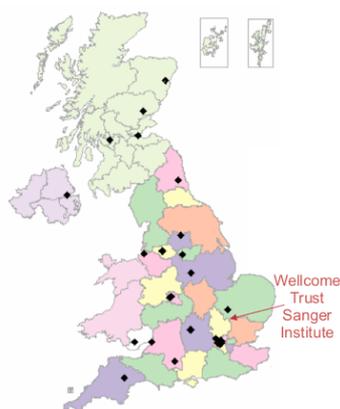
Welcome to the first edition of the DDD Family Newsletter!

This first newsletter coincides with release of the first set of DDD results to clinical teams. We'll be producing a Newsletter every Autumn until the end of the project in 2015, to keep you updated on the progress of the research and thank you for participating in DDD.

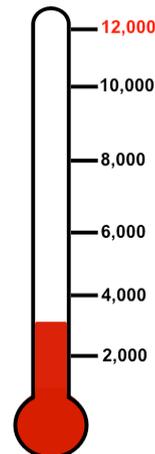
Deciphering Developmental Disorders is a pioneering study, and the largest of its kind in the world. Thanks to funding from the Wellcome Trust and the Department of Health, we are able to recruit patients throughout the entirety of the UK and give them access to the latest genetic technologies and scientific knowledge. Although not all the families in DDD will get a diagnosis for their child, many will, and the project as a whole will improve our understanding and management of developmental disorders and provide new avenues for research into treatments.

RECRUITMENT

All 23 NHS Regional Genetics Services across the UK are now recruiting families to DDD. We have a dedicated network of clinical geneticists and research nurses/genetic counsellors working towards finding and consenting eligible families, helping with taking samples, and collecting clinical information for DDD research. In addition to clinical teams throughout the country, we also have a dedicated team of scientists working on DDD at the Wellcome Trust Sanger Institute in Cambridge (where a third of the human genome sequence was first mapped). Here we all are at a recent meeting...



Since April 2011, we have recruited around 3,000 families into DDD, so we're well on the way to our target of 12,000. Thank you for being part of the study!



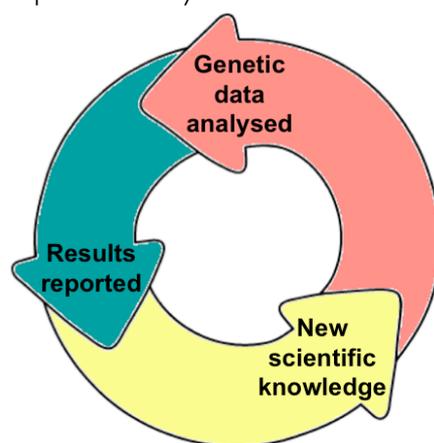


The primary objective of the DDD project is to improve clinical genetic practice for children with developmental disorders within a 5 year period by the systematic application of the latest microarray and sequencing methods to patients with disorders of development while addressing the new ethical challenges raised.

RESULTS

We are working hard to produce results that are both accurate and clinically relevant. Because of the enormous volume of genetic data, and the rapidly changing science of genomics, this inevitably takes time! However, the power of the DDD study lies in its size, so the more data we have, the better our chances of finding the cause of your child's developmental disorder.

We have now reported initial results for the first few families! Although some of you will receive results soon, many families will not have any results from our initial analysis. This may be because we have not yet completed all the genetic analyses for your child. Even if these are complete, and we have not found the cause of your child's difficulties, please be re-assured that we will continue to analyse and re-analyse all the genetic data periodically until the end of the study, so you may receive results later in the study. Your clinician will be alerted as soon as we report a new result so that they can re-contact you, check the result and discuss it with you. (We will also alert your clinician if we have finished an analysis but have not found a result yet, or if we need another sample of your child's DNA.)



SUPPORT GROUPS

Several patient support groups are actively involved with DDD. The first is **SWAN UK – Syndromes Without A Name** – a project run by **Genetic Alliance UK** offering support and information to families of children with undiagnosed genetic conditions (www.swanuk.wordpress.com). They are able to provide advice about living without a diagnosis, as well as answer specific questions relating to DDD.

The second patient support group involved with DDD is **Unique**, the rare chromosome disorder support group for families with a diagnosis of a chromosomal change that caused their child's developmental disorder (www.rarechromo.org).

Both groups have a thriving community of families, and you can find them on Facebook and Twitter!



KNOWLEDGE SHARING

We take great care to ensure that the data we generate is confidential and only shared through appropriate channels. Clinically relevant findings will be fed-back securely and directly to your clinical team for them to check and discuss with you. The enormous benefits of DDD can only be realised by sharing the knowledge gained as widely as possible with researchers working on developmental disorders. External researchers can only gain access to anonymised data in DDD if they have a valid research question relating to developmental disorders and commit to protecting the data. In addition to sharing data, we will also publish important findings and new discoveries resulting from DDD (all anonymised) in peer-reviewed journals so that other scientists, clinicians and patients around the world can benefit from the research. We intend to publish our first papers next year, and will keep an updated list of publications on www.ddduk.org. In addition, we have already started giving talks at national and international conferences about DDD, to inform researchers about the methods being developed in the project as well as important new findings. The study has also featured on TV already!

ETHICS RESEARCH

Despite the growing number of genetics research studies, there is no consensus on what information generated by the research is appropriate to share with research participants and indeed what participants would like to know. Although the policy on sharing data within DDD had to be decided at the outset, we are carrying out some ethics research to help inform the policies adopted in future genetic studies. If you're part of DDD, we want to know what you think! So please have a look at the videos on www.ddduk.org/ethicsresearch and fill in the questionnaire online – it's easy and fun to do, and anyone can participate.

Ethics and Genomics Survey reset answers & start again



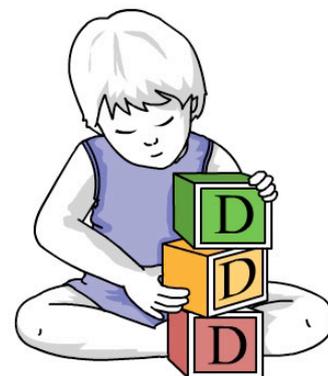
- ➔ Questions about you
- Sharing of Pertinent Findings
- Sharing of Incidental Findings
- Categorizing Incidental Findings
- Relations with Risk
- Raw data
- Duty of Genomic Researchers
- Filter of Genomic Information
- Consent for genomic research
- Last few questions about you

Questions about you

- Have you or your family ever been (or are currently) a research participant in a genetic research project?

Yes
 No
 Don't know

« Previous
Next »
Leave a comment



A Diagnosis

*We're pinning our hopes on
Deciphering Developmental
Delay*

*To provide some answers
come what may!*

*If one more doctor says a
diagnosis doesn't matter.*

*I think I might go mad as a
hatter!*

*We know it won't change
our special boy,*

*Making him better isn't our
ploy.*

*You don't understand why
we've made it our mission.*

*That's because you're not
in our position!*

With thanks to SWAN and
one of the families in DDD



Fascinating Facts

Everyone receives 3 billion letters of DNA from each parent.

About 1 in 1,000 of these is different between individuals.

Every child, healthy or not, has 50-100 new changes in their DNA.

Just a single genetic change can cause a developmental disorder...

For more information about DDD, please talk to your clinical geneticist, or visit our website for updates:
www.ddduk.org

TECHNOLOGIES

The first technology we use in the DDD study is called an **array**. This allows us to look across an individual's genome for copy number changes – gains or losses of part of a chromosome, which may affect many genes or just one. Although many people in DDD will already have received an NHS array, the DDD array is higher resolution so we are able to see smaller changes than most clinical tests.



The second technology we use in DDD is called **sequencing**. This allows us to determine the exact genetic code of all the genes, to look for small changes that might explain a child's disorder. This is a very difficult task because everyone has a very large genome with many harmless changes, and we are still developing new ways to look for disease-causing alterations. In addition, the price of sequencing is currently falling dramatically, so we are planning to do most of the sequencing towards the end of the project to get the most out of our funding. So you might not get a result from sequencing until near the end of the project.

FAQS

Q. What happens if we spat in the wrong tubes?

A. Don't worry, this is not a problem. We can work out which sample is which from the samples themselves.

Q. Will I have to give another sample in future?

A. Maybe. Because of the stringent quality control processes we use, some of the samples provided may not be appropriate for all our tests. If this is the case, we will let your clinical team know that we need another sample from your child. Also, because the tests are being done at a research institute, all the results will have to be checked in an NHS lab and you may need to give another sample for these validation tests.

Q. Will you tell us if you find something important in any of our family's genomes which doesn't relate to our child's developmental disorder?

A. No. We analyse the data specifically only looking for genetic changes that are likely to explain your child's developmental disorder. We filter the data extensively to remove as many other changes as possible, and we won't be searching for alterations associated with any other conditions (which are often termed 'incidental findings').