Why is research important in Dup15q syndrome?

Dup15q syndrome is a complicated set of core and associated symptoms resulting from the duplication of a portion of chromosome 15. Dup15q syndrome can affect people in many different ways, with some people finding certain symptoms harder to manage and therefore requiring additional support.

Existing medical interventions and behavioral therapies for symptoms of Dup15q syndrome can potentially improve development, function, and quality of life. It is important to note that there are a wide range of developmental disabilities experienced by individuals with Dup15q syndrome, meaning each person will require a different type of medical intervention.

Therefore, the Dup15q syndrome research community needs to do more research to understand which people with Dup15q syndrome will benefit from safe and effective medication and therapies. To do this, they conduct clinical trials.

What is a clinical trial?

New medications and therapies are tested through clinical trials (also known as research or clinical studies). These involve volunteers who trial the new medication or therapy, usually compared against already available medication, therapy, or medication with no effect, known as a placebo.

Participants of trials receive a new medication or therapy and are closely assessed by a team of trained doctors, nurses, and research staff to see its effects. Data are collected from all the participants, which allow investigators (those running the trial) to accurately determine whether the medication or therapy is safe and effective.

Why do clinical trials matter to me, my family, and the Dup15q syndrome community?

Clinical trials have enabled the wider scientific community to discover many different and essential medicines, like painkillers, antibiotics, and vaccines that we have today. This also means that the Dup15q syndrome research community may find new interventions and therapies as their understanding of Dup15q syndrome improves.

In Dup15q syndrome, clinical trials aim to:

- **Understand** more about Dup15q syndrome and how it affects people differently
- **Appreciate** how addressing certain associated Dup15q syndrome symptoms can be beneficial for people with Dup15q syndrome
- **Discover** medication and therapies that are effective, safe and work alongside other interventions
- **Improve** the lives of people with Dup15q syndrome, their families, and supporters
The clinical trial process:

Every medication or therapy, including those for Dup15q syndrome, goes through various Phases of clinical trials, each with different objectives, before it can be approved for use by health authorities. Data found at each Phase determines whether research continues. Once the medication or therapy is approved and available, it will continue to be monitored for many years to investigate any potential long-term side-effects.

The diagram below describes a typical medication or therapy’s journey through a successful clinical trial process:

Key clinical trial terms

There are many different terms you may hear during a Dup15q syndrome clinical trial. The following terminology explains some key words that you are likely to encounter regarding types of clinical trials and how they will be conducted.

<table>
<thead>
<tr>
<th>Term</th>
<th>Description</th>
</tr>
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<tbody>
<tr>
<td>Standard of care</td>
<td>The medication or therapy that is already available outside of the trial. At the time of print, no medication is available for core Dup15q syndrome symptoms.</td>
</tr>
<tr>
<td>Placebo</td>
<td>A substance that has no effect but is designed to look the same as the trial medication or therapy and may be given to some participants. Placebos are vital in clinical trials to show if an actual medication or therapy works.</td>
</tr>
<tr>
<td>Placebo effect</td>
<td>A placebo effect is when a person's physical or mental health appears to improve after taking a placebo or 'dummy' treatment.</td>
</tr>
<tr>
<td>Comparative trial</td>
<td>A type of study where one group receive the trial medication and other groups receive either the standard of care or placebo to compare how safe and effective the trial medication or therapy is.</td>
</tr>
<tr>
<td>Randomised</td>
<td>Participants may be put into different groups, without knowing which, at the start of the trial. For example, one group could be given a certain dose of the trial medication and the other group could receive a different dose or a placebo.</td>
</tr>
<tr>
<td>Double-blind</td>
<td>Neither the investigators or the participants know which group is receiving the medication or placebo until after the trial finishes.</td>
</tr>
<tr>
<td>Pseudonymised data</td>
<td>Data collected throughout the trial cannot be linked back to the individual participating. All clinical trials will ensure that data is pseudonymised, meaning any data that could identify a participant is replaced with other values. For example, someone's name may be replaced as Participant 1.</td>
</tr>
<tr>
<td>Open label extension</td>
<td>Once the trial finishes, all participants, regardless of whether they received the trial medication or placebo, are invited to take the trial medication for a further period to gain further data on how the drug works and how safe it is. This is sometimes also referred to as a long term extension study. Not all trials will offer an open label extension.</td>
</tr>
</tbody>
</table>
What should my family and I expect to happen during a clinical trial?

How a clinical trial is conducted will vary depending on factors like the Phase and location. However, you can usually expect the following stages to occur (both you and your child will need to be involved in most stages).

Recruitment
You may be approached by your child's doctor or a community organisation who think a clinical trial may be of interest to you and will describe it to you.

Pre-screening
You answer a set of questions to help the trial organisers understand more about your child and whether the trial will be suitable. These are typically done over the phone or online, but can also be done in person.

Screening
You will travel to the trial site, meet the staff, and ask any questions you may have. Your child will then undergo further physical tests, questionnaires, and possibly laboratory testing (e.g. blood or urine tests) or scans (e.g. MRI). Based on the results, your child may or may not qualify to participate in the rest of the trial.

Informed consent
You are given all the facts about the trial, such as what the medication or therapy is, any expected side effects, the tests that your child will be a part of etc. You can then decide whether you wish for your child to participate by choosing to sign an informed consent form.

Enrolment and group assignment
If your child is selected for the trial, you will be assigned into different groups (often randomly) to receive the trial medication or therapy, standard of care, or a placebo, depending on the trial.

Study visits and monitoring
You and your child will attend the trial site and receive different types of tests and questionnaires. You can ask any questions or raise concerns. (Your child may also be visited by trial staff, fill out questionnaires, or wear measuring devices at home, meaning they will not need to visit the trial site).

Analysis and completion
Your child's data, along with data from the other participants, are reviewed, analysed, and interpreted. The results are presented at scientific conferences, published online in scientific journals, and made available to participants, supporters, and the public (there is sometimes a cost to access).

End of study
The trial ends and your child may stop receiving the trial medication, therapy, or placebo. If there is an open-label extension, your child may be invited to continue or start receiving the trial medication or therapy.

Clinical trials are conducted under very strict safety controls, and your child will be regularly monitored to make sure they are well and healthy throughout.

Your child can leave the trial at any point for whatever reason, even after signing the informed consent form.
What will happen with the data?

Your child’s data is absolutely essential to achieve the aims of the clinical trial. The infographic below explains why.

Data are collected continuously throughout the trial in many different forms, such as your child’s results in tests, questionnaires, blood samples, vital signs like pulse rate and blood pressure, brain activity, and DNA.

As part of the informed consent process, you should be clearly told how your child’s data will be used and who will have access to it. Please raise any data concerns you may have with the trial organisers as early as possible.

What are the advantages and disadvantages for me and my child in joining a clinical trial?

You and your child should take some time before deciding to join a clinical trial. While there are several positives of participating, there are also a number of potential negatives.

<table>
<thead>
<tr>
<th>The possible advantages of joining a clinical trial are:</th>
<th>The possible disadvantages of joining a clinical trial are:</th>
</tr>
</thead>
<tbody>
<tr>
<td>receiving a medication or therapy that improves your child’s life more than what’s already available</td>
<td>receiving a medication or therapy that is no better or worse than what’s already available, including a placebo</td>
</tr>
<tr>
<td>receiving the best available monitoring and care and more opportunities to ask questions</td>
<td>your child may need to stop receiving a medication or therapy that they currently take</td>
</tr>
<tr>
<td>free care during the trial</td>
<td>having side effects</td>
</tr>
<tr>
<td>contributing to important research that can help you and other Dup15q syndrome families in the future.</td>
<td>having tests that can be uncomfortable or scary</td>
</tr>
<tr>
<td></td>
<td>time and travel commitments.</td>
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</tbody>
</table>

What important questions should I ask my child's doctor about clinical trials?

If your child is interested in participating in a clinical trial, you should ask your doctor:

Are there any clinical trials that my child can join?

If your doctor offers your child a place on a clinical trial, you may want to ask the following questions.

- What is the purpose of the trial?
- Will my child have to stop taking any medication that they already take?
- How long will the trial last?
- What if my child wants to leave the trial?
- What is the treatment or therapy that you are investigating?
- Who can I speak to if myself or my child have further questions?
- What are the treatment or therapy’s side effects?
Who should my child and I speak to if we want more information on clinical trials?

Speak to your doctor or local Dup15q syndrome community organisation (if available) for more information on available clinical trials or if you have any questions about joining a trial.

Websites like ClinicalTrials.gov contain all current and future studies in Dup15q syndrome, as well as trial details and results.

The following online videos explain the clinical trial process further.

**Clinical Trials**
https://www.youtube.com/watch?v=5zXuON7Rueo

**What is a Clinical Research Study?**
https://www.youtube.com/watch?v=KCmfCWHM15a

**The Importance of Clinical Trials**
https://www.youtube.com/watch?v=15hknL5m6W0

**Designing a Clinical Trial**
https://www.youtube.com/watch?v=vV6UzbZP3IU

**The Journey of Clinical Trials**
https://www.youtube.com/watch?v=ru-2mKxhpM54

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