

FACEMATCH INFORMATION

Research Project Title: The FaceMatch Project (**HREC project number:** 17/05/17/4.06)

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We would like to invite you to participate in **The FaceMatch: Searching for a Diagnosis** project.

WHAT IS THE BACKGROUND TO THIS RESEARCH PROJECT?

Parents of a child with intellectual disability (ID) often spend many years and medical appointments searching for a diagnosis. Although there are various prenatal and environmental causes of intellectual disability (ID), the most common cause is genetic. Genetic changes can be inherited from one or both parents, but often occur for the first time in a child. Understanding the genetic cause of ID can guide medical management and reduce the sense of isolation for the family.

Improved genetic testing has made it possible to test an individual's entire genetic makeup (the genome) including all the known (approximately 1500) genes important for brain development, in a single test. However, a diagnosis is still only achieved approximately 50% of the time. There are a large number of ID genes still to be discovered. Understanding the genes involved in brain growth and function is the first step towards developing treatments in the future.

WHAT IS THE AIM OF THE FACEMATCH PROJECT?

FaceMatch aims to suggest a diagnosis in people where genetic testing has not provided an answer. People with the same genetic condition can often share similar facial features. By matching facial images and written descriptions of undiagnosed children from around the world, the project aims to provide a diagnosis for more families and to discover new ID genes. As advanced genetic testing is not available for all families, we will also use the technology to match the faces of undiagnosed people with those who have a confirmed genetic diagnosis.

WHO CAN PARTICIPATE IN THE FACEMATCH PROJECT?

- 1) Parents of children or adults with ID who remain without a genetic diagnosis following a review by a clinical geneticist.
- 2) We would be grateful if parents of children or adults with ID who have a confirmed genetic diagnosis would consider contributing photographs to FaceMatch. This will improve our image database and help other families still searching for a diagnosis.

WHAT DOES PARTICIPATION IN THE FACEMATCH PROJECT INVOLVE?

You or the clinical geneticist or medical specialist nominated by you, will be asked to provide information for your son or daughter's profile page in the secure FaceMatch database. This will include uploading photographs of them at different ages and providing relevant medical information.

Every new image uploaded into the FaceMatch database will be matched against all other photographs within the database. Photographs identified as very similar will be reviewed by the FaceMatch team and the doctors of the matching individuals, within the FaceMatch project. Your nominated doctor will contact you if a high probability facial match is made.

WHAT ARE THE POSSIBLE BENEFITS OF TAKING PART IN THIS PROJECT?

The main benefit of this project is the potential to help families find a diagnosis.

WHAT ARE THE POSSIBLE RISKS OF TAKING PART IN THIS PROJECT?

It is possible that a facial match may not lead to a diagnosis. This could generate concern or disappointment for you. It is also possible that participation in the project may lead to the diagnosis of an inherited form of intellectual disability. This information may have implications for you, your children or other family members.

Even when parents have been searching for a diagnosis to explain the intellectual disability in their child, the actual time of diagnosis can be distressing. Support from your nominated doctor and/or a genetic counsellor will be available.

WHAT WILL BE DONE TO ENSURE THAT INFORMATION GIVEN TO FACEMATCH IS CONFIDENTIAL?

Any information collected for this research project will be treated as confidential. Information and photographs are stored securely away from the publically viewed FaceMatch website.

All uploaded photographs and data will be encrypted and stored behind a firewall in a password protected computer at the Hunter Medical Research Institute. We will remove your name and your son/daughter's name and give the information a special code. Only the research team can match the names to their code.

De-identified data generated by the computer vision software from the photograph is used for matching purposes. When a facial match is made, images can only be viewed within the secure FaceMatch site by the FaceMatch team and the nominated doctors of the matching participants. If a match is confirmed and parents of both matching individuals have consented to image sharing, parents will be able to view matching images.

When we write or talk about the results of this project, information will be provided in such a way that people in your family cannot be identified.

WHAT IF YOU CHANGE YOUR MIND?

Participation in this research project is voluntary. It is your choice to let your son or daughter take part in the project. You do not need to agree to participate if you do not want to. If you give your consent and then change your mind, you can withdraw from the project at any time by emailing our team at info@FaceMatch.org.au. You do not need to tell us the reason why you want to withdraw.

WILL YOU BE INFORMED OF FACEMATCHING RESULTS?

If your son or daughter has a facial match with another individual within the database, information about this match will be sent to your nominated doctor who will contact you.

Thank you for taking the time to read this information statement.

If you would like more information about the project or if you need to speak to a member of the research team please contact the FaceMatch team at info@FaceMatch.org.au

If you have any concerns and/or complaints about the project, the way it is being conducted or your child's rights as a research participant, and would like to speak to someone independent of the project, please contact Nicole Gerrand, Manager, Research Ethics and Governance, Hunter New England Health Research Ethics committee on HNELHD-HREC@hnehealth.nsw.gov.au. Please quote reference number 17/05/17/4.06