MassGeneral Hospital for Children | MASSACHUSETTS GENERAL HOSPITAL | HARVARD MEDICAL SCHOOL

GEMMA

Multi-Omics Research in Autism

Genome, **E**nvironment, **M**icrobiome and **M**etabolome in **A**utism

GEMMA Study

Do you have a child diagnosed with autism spectrum disorder (ASD)?

Are you pregnant or planning to get pregnant, and wondering what the risk of ASD will be in your newborn?

What is the goal of this study? To understand the role that our genes, gut microbiome, and environmental factors play in the development of autism. To identify potential biomarkers (in the blood, stool, urine and saliva) predictive of autism development and ultimately predict, prevent and treat ASD symptoms and GI comorbidities.

Who can participate? Infants 0-6 months of age who have a sibling diagnosed with ASD.

What is involved?

- Periodic collection of blood, stool, urine and saliva samples over a 3-year period
- Periodic questionnaires regarding child's social, medical and dietary history
- Autism evaluation every 6 months starting at 1 year of age

Children who develop autism while enrolled will be eligible for the GEMMA interventional study: This study will use the identified biomarkers to implement personalized prevention and treatment of ASD and ASD-related symptoms.

For more information, contact us at

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Massachusetts General Hospital
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MassGeneral Hospital for Children

Genome Environment Microbiome and Metabolome in Autism

The MGHfC Mucosal Immunology and Biology Research Center has launched a groundbreaking research project to better understand the many factors that contribute to the development of autism spectrum disorders. MGHfC is one of 16 international partners in this multi-center European Commission project. Read this handout to learn how you can get involved.

WHAT IS THE GOAL OF THE STUDY?

The **G**enome, **E**nvironment, **M**icrobiome and **M**etabolome in **A**utism (GEMMA) Study is a multi-center observational study that follows children who are genetically at risk of developing autism for their first 3 years of life. During this time, we will explore if there is a relationship between the bacteria in the gut (microbiome) and the development of autism, as well as the role that genes and environmental factors may play. We are hopeful that this study will help us to better understand why some children develop autism while others do not, and ultimately to predict autism development in children prior to its onset.

WHO CAN PARTICIPATE?

- Infants 0-6 months of age who have a biological sibling diagnosed with autism.
- If you are currently pregnant or planning to get pregnant and you have a child with autism, you can begin the enrollment process before birth!

WHAT ARE THE BENEFITS OF PARTICIPATING?

Your child's development will be monitored throughout the study, and autism assessments will be done every 6 months starting at 1 year of age. If your child develops autism while enrolled in the study, he or she will not only benefit from early diagnosis but will also have the opportunity to enroll in the GEMMA interventional study – which will use the identified biomarkers to implement personalized prevention and treatment of ASD symptoms and GI comorbidities.

Participants have a first-hand, active role in groundbreaking science and around-the-clock access to the GEMMA study team, comprised of experts in the field of autism spectrum disorders. Our hope is that the information we learn from this study may lead to earlier diagnosis than is currently possible, and possible solutions alleviating and/or preventing ASD and ASD-related symptoms in patients in the future.

WHAT DOES PARTICIPATION INVOLVE?

- We will collect blood, stool, urine and saliva samples every 6 months until the age of 3. Apart from blood, samples can be collected at home and mailed back to the study site. Blood draws can be completed at the study site or at your child's pediatrician's office / a local lab (and mailed back to the study site).
- You will answer monthly questionnaires regarding your infant's development, as well as their medical and dietary history. All surveys can be completed securely online or on paper if preferred.
- Your child will complete an autism assessment with a qualified staff member every 6 months starting at 1 year of age.

HOW CAN I LEARN MORE?

Visit our website at https://www.gemma-project.eu/.

Email us at MGHGEMMA@mgh.harvard.edu to learn more or enroll.

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