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Spit samples uncover genetic risk factors for pediatric obsessive-compulsive disorder

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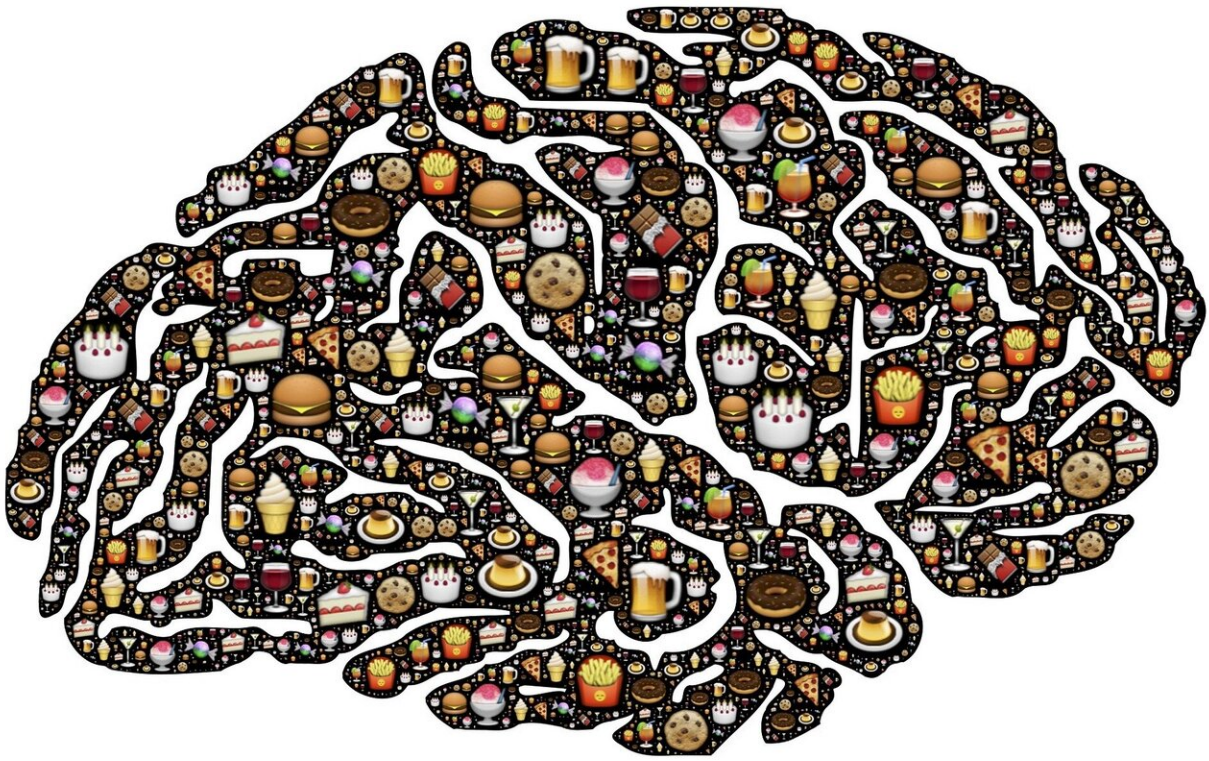

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Researchers at the University of Calgary and The Hospital for Sick

Children (SickKids), in Toronto, have discovered genetic risk factors for OCD that could help pave the way for earlier diagnosis and improved treatment for children and youth.

"Our group made the first finding of a genome-wide significant risk gene relevant to childhood OCD," says Dr. Paul Arnold, MD, Ph.D., co-principal investigator, a professor and director of The Mathison Centre for Mental Health Research & Education at the Cumming School of Medicine. "We've known that OCD runs in families, but we hadn't identified and validated specific genetic risks of OCD symptoms in children and youth until now."

The research drew on the Spit for Science study, a research project led by SickKids looking at how genes interact with the environment to impact physical and mental health. Participants from the community were recruited via an innovative research design run out of the Ontario Science Centre, which has generated a diverse sample of 23,000 participants thus far. Participants provide a DNA sample through their saliva, do a cognitive task, and complete questionnaires on their health, lifestyle and behaviors.

In this study, saliva samples from over 5,000 children and youth were scanned and compared to participant responses using the Toronto Obsessive-Compulsive Scale (TOCS). The TOCS is a questionnaire used to evaluate obsessive-compulsive traits developed by Dr. Arnold and the team at SickKids. After looking across millions of genetic variants from the saliva samples, the team identified that children and youth with a genetic variant in the gene PTPRD had a greater risk for more obsessive-compulsive traits. The findings are published in *Translational Psychiatry*.

"Discovering the genes involved in OCD is critical to help improve patients' lives. It is still early days, but our hope is these findings will lead us to understand the causes of OCD, which in turn could help

identify people with OCD sooner and develop better treatments," says Dr. Christie Burton, Ph.D., lead author and research associate in the Neurosciences & Mental Health program at SickKids.

The research team, which also includes co-principal investigators, Drs. Jennifer Crosbie, Ph.D., Clinical Psychologist at SickKids, and Russell Schachar, MD, Psychiatrist at SickKids, highlight that a greater understanding of the underlying genetics may eventually be an important complement to clinical assessment and could help guide treatment options in the future.

"OCD can present very differently and at various ages in each individual, adding to the challenge of treatment and diagnosis," says Crosbie, who is also an associate scientist in the Neurosciences & Mental Health program at SickKids. "Studies like this one are an important step towards developing precision medicine approaches for mental health."

Sam, 17, lives with OCD and with therapy and medication, he says he has been able to face his obsessions and compulsions, ride out the anxiety and control his actions. Looking back at his childhood, Sam says he had some OCD tendencies as early as elementary school, but neither he nor his family realized he had a mental illness. The researchers hope that by understanding the genetics of OCD they can develop better treatments, improve outcomes and diagnose youth like Sam earlier.

"At first I wasn't sure what to do with the diagnosis, it was very foreign, I didn't want to perceive myself as having a mental health issue," says Sam. "But, knowing I have OCD helped me overcome the challenges. With therapy and medication, I've stopped OCD from overtaking my life and taken back control."

Sam is a real teenager, but Sam isn't his real name. He says due to the stigma around OCD he would prefer to remain anonymous.

More information: et al, Genome-wide association study of pediatric obsessive-compulsive traits: shared genetic risk between traits and disorder, *Translational Psychiatry* (2021). DOI: 10.1038/s41398-020-01121-9