

Autism Speaks Through Gene Expression

Philadelphia, Pa. – Autism spectrum disorders affect nearly 1 in 88 children, with symptoms ranging from mild personality traits to severe intellectual disability and seizures. Understanding the altered genetic pathways is critical for diagnosis and treatment. New work to examine which genes are responsible for autism disorders will be presented at the 57th Annual Meeting of the Biophysical Society (BPS), held Feb. 2-6, 2013, in Philadelphia, Pa.

“Autism is the most inheritable of neurodevelopmental disorders,” explains Rajini Rao of Johns Hopkins University in Baltimore, Md., “but identifying the underlying genes is difficult since no single gene contributes more than a tiny fraction of autism cases.” Rather, she continues, “mutations in many different genes variably affect a few common pathways.”

A team of scientists at Johns Hopkins and Tel Aviv University in Israel looked at genetic variations in DNA sequence in the ion transporter NHE9 and found that autism-associated variants in NHE9 result in a profound loss of transporter function. “Altering levels of this transporter at the synapse may modulate critical proteins on the cell surface that bring in nutrients or neurotransmitters such as glutamate,” says Rao. “Elevated glutamate levels are known to trigger seizures, possibly explaining why autistic patients with mutations in these ion transporters also have seizures.”

A unique aspect of the team’s approach was that they exploited decades of basic research done in bacteria and yeast to study a complex human neurological disorder. First, the group at Tel Aviv University, led by Nir Ben-Tal, built structural models of NHE9 using a bacterial relative as a template, allowing the Rao laboratory at Johns Hopkins to use the simple baker’s yeast for screening the mutations. In the future, as genomic information becomes readily available for everyone, such easy, inexpensive, and rapid screening methods will be essential to evaluate rare genetic variants in autism and other disorders.

Rao and her team are optimistic about the potential benefits of their latest findings. “Although the research is still at an early stage, drugs that target the cellular pathways regulated by NHE9 could compensate for its loss of function and lead to potential therapy in the future,” Rao says. “These findings add a new candidate for genetic screening of at-risk patients that may lead to better diagnosis or treatment of autism.”

Presentation #118-Plat, “Functional evaluation of autism associated mutations in SLC9A9 (NHE9),” will take place at 9:15 a.m. on Sunday, Feb. 3, 2013, in the Pennsylvania Convention Center, Room 113C. ABSTRACT: <http://tinyurl.com/apjmm7a>

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This news release was prepared for the Biophysical Society (BPS) by the American Institute of Physics (AIP).

ABOUT THE 2013 ANNUAL MEETING

Each year, the Biophysical Society Annual Meeting brings together over 6,000 research scientists in the multidisciplinary fields representing biophysics. With more than 3,900 poster presentations, over 200 exhibits, and more than 20 symposia, the Annual Meeting is the largest meeting of biophysicists in the world. Despite its size, the meeting retains its small-meeting flavor through its subgroup meetings, platform sessions, social activities, and committee programs.

The 57th Annual Meeting will be held at the Pennsylvania Convention Center (1101 Arch Street, Philadelphia, PA 19107). For maps and directions, please visit:

<http://www.paconvention.com/explore-philadelphia/directions-and-parking>.

QUICK LINKS

Meeting Home Page:

<http://www.biophysics.org/2013meeting/Main/tabid/3523/Default.aspx>

Housing and Travel Information:

<http://www.biophysics.org/2013meeting/AccommodationsTravel/HotelInformation/tabid/3621/Default.aspx>

Program Abstracts and Itinerary Planner:

<http://www.abstractsonline.com/plan/start.aspx?mkey=%7B763246BB-EBE4-430F-9545-81BC84D0C68C%7D>

PRESS REGISTRATION

The Biophysical Society invites credentialed journalists, freelance reporters working on assignment, and public information officers to attend its Annual Meeting free of charge. For more information on registering as a member of the press, contact BPS Director of Public Affairs and Communications Ellen Weiss at eweiss@biophysics.org or 240-290-5606, or visit <http://www.biophysics.org/2013meeting/Registration/Press/tabid/3619/Default.aspx>. Press registration will also be available onsite at the Pennsylvania Convention Center in the Biophysical Society's meeting office, Room 304VIP.

ABOUT BPS

The Biophysical Society (BPS), founded in 1958, is a professional scientific society established to encourage development and dissemination of knowledge in biophysics. The Society promotes growth in this expanding field through its annual meeting, monthly journal, and committee and outreach activities. Its 9000 members are located throughout the U.S. and the world, where they teach and conduct research in colleges, universities, laboratories, government agencies, and industry. For more information on the Society or the 2013 Annual Meeting, visit www.biophysics.org.

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