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Knowledge gap in genetic testing for autism

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One of every 88 children born in the United States will be diagnosed with a form of autism, according to the US Centers for Disease Control and Prevention. Genetic testing for autism spectrum disorders (ASDs) has been available for a decade. However, the current rate of identifying an underlying genetic cause is less than 25%. In addition, genetic testing for ASDs raises issues of potential genetic discrimination, privacy concerns, and a potential psychological burden on parents and families. To assess current understanding and awareness of ASD genetic testing, Chen et al. conducted structured interviews with 42 parents of children diagnosed with autism. The research team recruited parents from community-based autism support groups in Texas to obtain a diverse ethnic and socioeconomic sample. Twelve parents in the sample had taken their child or children for genetic testing. Of the remainder, 63% had never heard of genetic testing for autism prior to the interview. Most of the parents (29 of 42) held positive attitudes about genetic testing, but 11 had negative attitudes, citing a variety of reasons. Most parents had learned about genetic testing through the mass media or scientific articles; only two interviewees had learned about ASD genetic testing from their children's doctors. The authors advocate more education about autism genetic testing for both health-care providers and the public, as well as additional research to assess the influence of ethnic and socioeconomic status on attitudes toward genetic testing for autism. *—Karyn Hede, News Editor*

Single-gene defect, complex disease: explaining the disconnect

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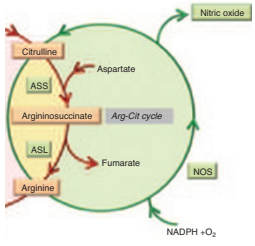

Ayelet Erez, winner of the 2012 William K. Bowes, Jr. Award in Medical Genetics, presents a review of complex phenotypes observed in inherited metabolic disorders, using the case of argininosuccinic aciduria (ASA) as a prototype. Erez, until recently a medical geneticist at Texas Children's Hospital in Houston, has joined Israel's Weizmann Institute, where she plans to establish a pediatric cancer genetic clinic. Her research has focused on understanding ASA, a rare inherited disorder caused by a defective gene necessary to make the enzyme argininosuccinate lyase (ASS1). Without the enzyme, the body can't synthesize arginine, an essential amino acid. Her work using a mouse model of the disease helped establish that this single gene participated in more biological pathways than had previously been appreciated, including production of nitric oxide (NO), a known vasodilator. Patients with ASA can have chronic hypertension as a result. In her review, Erez shows how a single-gene defect can alter multiple metabolic pathways, leading to complex symptoms and predisposition to disease states. For example, although ASS1 contributes to the urea cycle in the liver, it is also expressed in other tissues, driven by the need for arginine in other metabolic pathways, including production of NO. Erez provides anecdotal evidence of a role for NO supplementation in treatment of some ASA patients and calls for a more nuanced understanding of interrelated metabolic pathways, with the goal of developing new treatments for inherited metabolic diseases. *—Karyn Hede, News Editor*

NEWS BRIEFS

Girls may be protected from autistic behavior


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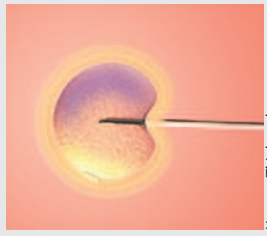
A study of twin pairs in the United Kingdom and Sweden recently revealed a female protective effect from autistic impairment, providing an explanation for the well-documented sex differences in autism diagnosis. Given that autistic behavior often runs in families, the authors hypothesized that, if a protective effect exists, siblings of girls with high autistic-behavior scores should carry more risk factors than the siblings of autistic boys. Corresponding author Elise Robinson of Harvard Medical School and an international research team tested their theory in two large independent cohorts of fraternal twins: 3,842 twin pairs in a UK-based twin study and 6,040 twin pairs in a Swedish twin study. After identifying pairs in which one twin scored in the top 10% of autistic impairment, the authors compared the sibling impairment. They found that siblings of girls indeed displayed greater average impairments than the siblings of boys, a finding published 18 February 2013 in the *Proceedings of the National Academy of Sciences*. The authors concluded that boys require fewer familial risk factors to have an equivalent impairment, a finding that could have implications for the design and interpretation of future autism genetic association studies.
—Karyn Hede, News Editor

UK gene review draws ire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A move to review the appropriateness of some genetic testing prior to embryo implantation in the United Kingdom has raised questions about whether improved treatments for genetic diseases should be taken into account when parents request testing. In the United Kingdom, preimplantation genetic diagnosis (PGD) is strictly regulated through the Human Fertilisation Embryology Authority. Since 1991, it has operated as a government-authorized independent regulator that licenses fertility clinics carrying out in vitro fertilization, artificial insemination, and human-embryo research. Now a move to review seven previously approved conditions for which medical treatment is thought to have improved since the conditions were first licensed has ignited debate. In a commentary

NEWS BRIEFS

recently published in the UK-based *BioNews*, a group of reproductive-medicine experts question whether an attempt to prohibit PGD for inherited disorders implies that it should also no longer be offered in the context of prenatal testing. The authors point out that the potential seriousness of a condition can only be estimated, because many nongenetic factors can influence the severity of an inherited disorder. Clearly, as both genetic diagnosis and therapeutic interventions improve, such decisions will become both medically and ethically trickier.
—Karyn Hede, News Editor

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Genetics in Medicine is a monthly journal committed to the timely publication of:

- Original reports which enhance the knowledge and practice of medical genetics
- Strategies and innovative approaches to the education of medical providers at all levels in the realm of genetics

As the official journal of the American College of Medical Genetics and Genomics (ACMG), the journal will:

- Provide a forum for discussion, debate and innovation concerning the changing and expanding role of medical genetics within the broader context of medicine
- Fulfill our responsibility to the College membership through the publication of guidelines, policy statements and other information that enhances the practice and understanding of medical genetics

Finally, as genetics becomes increasingly important in the wider medical arena, we will be an accessible and authoritative resource for the dissemination of medical genetic knowledge to providers outside of the genetics community through appropriate reviews, discussions, recommendations and guidelines.