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Parents should know "savior baby" not realistic in most cases

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When a child is diagnosed with Fanconi anemia (FA), a rare genetic syndrome that results in bone marrow failure and a predisposition to cancer, the only option for treatment has been a genetically matched stem cell transplant. When neither parent is a genetic match, some families opt to try to conceive a "savior" sibling using in vitro fertilization (IVF) of a genetically matched embryo. But little information about the chances of success using this technique has been available. Now a research team from Barcelona, Spain, reports only one baby born after 38 IVF attempts by seven families over 11 years. To avoid bias, the researchers followed all families at their clinic attempting the procedure. Of the 299 embryos generated by IVF, only 75 had the proper genetic match, and only a fraction of these were candidates for implantation. A total of 17 transferred embryos resulted in five pregnancies but only one birth, for a success rate of 2.6%. The average age of women attempting the procedure was close to 40, given that children are generally not diagnosed with FA until age 7. The researchers suggest that advanced maternal age combined with an already low statistical chance of identifying a matched embryo make conceiving a "savior baby" unrealistic. Parents should be informed of these low chances, the authors state, given the emotional and economic costs of IVF. However, recent scientific and medical advances in transplantation and gene therapy may someday offer families superior treatment options. *—Karyn Hede, News Editor*

First diagnosis of Duchenne muscular dystrophy via a noninvasive prenatal test

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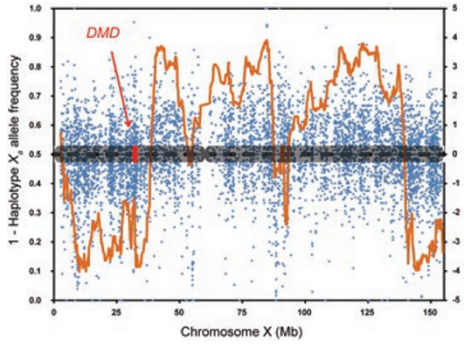

A research team based in Shanghai, China, has for the first time successfully diagnosed the most common form of muscular dystrophy using noninvasive prenatal testing (NIPT). Duchenne muscular dystrophy (DMD) is inherited via the X chromosome and affects 1 in 3,600 to 6,000 boys. The researchers used a technique they developed to recover, in a single step, the fetal genotype and both parental sequences from DNA in maternal blood plasma. Using this approach, they accurately predicted the mutation status of the fetus in eight pregnant women known to be DMD carriers. The tests showed that three of the fetuses were girls, two of whom did not inherit the DMD mutation. All five of the male fetuses were positive for the DMD trait. The results were confirmed by amniocentesis. Prior to this study, the researchers had shown that the technique effectively identified several autosomal recessive disorders, including congenital adrenal hyperplasia, maple syrup urine disease, and congenital deafness. However, no one had used NIPT to noninvasively predict the fetal mutation status in an X-linked disease. The test is not yet ready for routine clinical use, but the ability to diagnose common genetic disorders noninvasively early in pregnancy is sure to have important implications, both medical and ethical. *—Karyn Hede, News Editor*

NEWS BRIEFS

Beginning of the end for ethically neutral genetic counseling?

The official position of the National Society of Genetic Counselors (NSGC) is that "reproductive decisions should be made in the context of unbiased and comprehensive information, free from discrimination or coercion." But lawmakers in a number of US states have begun to chip away at the core principle of ethically neutral genetic counseling, enacting laws

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that require counselors to impart information mandated by legislative action. Eight states have now passed versions of the Down Syndrome Information Act into law. The legislation requires that specific information provided by the state be given to parents upon a diagnosis of Down syndrome, the most common chromosomal abnormality, affecting about 1 in 800 live births. In at least one state—Pennsylvania—the official state information resource contains information counter to NSGC best practices (http://www.health.pa.gov/My%20Health/Diseases%20and%20Conditions/A-D/Documents/eng.pdf). In response, the NSGC created its own information sheet (http://nsgc.org/d/do/4640) and has encouraged states to use

NEWS BRIEFS *(continued)*

its resources. But such legislation could be opening the door to attempts to legislate the information patients receive about other genetic conditions. The future autonomy of patients and genetic counselors is discussed in an opinion piece by bioethicist Arthur Caplan of New York University Langone Medical Center in a recent PLOS Biology article. *—Karyn Hede, News Editor*

Seemingly unrelated autoimmune disorders share genomic pathways

A new genome-wide association study (GWAS) has identified a network of shared regulatory pathways among 10 pediatric-onset autoimmune diseases. The study, published by *Nature Medicine* on 24 August 2015, pinpoints shared signals among disorders that were not previously understood to have a common origin. The authors state that it is the first systematic examination of shared genetic risk looking simultaneously at the genotype level across multiple autoimmune diseases. The results point to signaling pathways involved in cytokine antigen processing and presentation as well as helper T-cell activation. The study, which looked in detail at 27 genetic loci, showed that more than 70% (19) of the 27 were shared by at least three autoimmune diseases. Some of the associations had been previously reported, but others were newly discovered. The implied genetic sharing of pathways among autoimmune diseases suggests targeted therapeutic interventions that could benefit patients across several of the related disease states, the research team suggests. Many of the shared risk variants affect genes that have multiple biological roles and are already targets for clinical intervention in other diseases. However, because the study included only individuals of self-reported European ancestry, application of the findings could be limited in scope and, of course, must be replicated in future studies, given the history of seemingly promising GWAS failing to hold up to further investigative scrutiny. *—Karyn Hede, News Editor*