flourishing families™ 2019

10th Annual • FREE

SNRFSD.org

SPECIALNEEDS RESOURCE FOUNDATION OF SAN DIEGO

1,050+ RESOURCES FOR FAMILIES WITH SPECIAL NEEDS

ROLLING WITH IT

LOCAL ATHLETES PROVE LIFE ON WHEELS HAS NO BOUNDARIES

NAVIGATING MENTAL HEALTH

HOW TO GET HELP FOR YOUR CHILD

Published by SAN DIEGO family

Lisa Salz, MS, LCGC

What to know about Genetic Testing

Benefits, risks and limitations

People consider genetic testing for many reasons, such as having a child with special needs, having a family history of a disorder, planning for pregnancy or simple curiosity. A genetic diagnosis can help put pieces of a puzzle together, as in the case of this family:

Karen had an uneventful pregnancy with her daughter Eva, just as she had with two older children. Karen was in excellent health and took care to avoid unnecessary exposures during pregnancy; prenatal screenings indicated the pregnancy was low-risk. Eva arrived three weeks early, but Karen wasn't worried until she didn't hear crying after delivery. In fact, Eva was limp and quiet. She had unusual physical features, including a smaller head (microcephaly) and a high arched palate that made it difficult to latch on for breastfeeding. Eva also had a congenital heart defect that required surgery at 4 months of age. Eva's parents couldn't help but wonder why. They saw many specialists, but none could provide a reason for Eva's health issues and developmental delay.

The family was referred to a medical geneticist when Eva was almost one. After a thorough physical examination, the geneticist suspected Eva had Kabuki syndrome, a rare genetic condition with characteristic facial features that can affect multiple parts of the body. Genetic testing confirmed the diagnosis—a damaging variant in the KMTD2 gene was found. The geneticist and a genetic counselor reviewed the inheritance of Kabuki syndrome (and its genetic basis) with Eva's parents, which provided a reason for Eva's health issues. The diagnosis was not their fault. Eva's parents found other families who have children with Kabuki syndrome who offered practical information and much-needed emotional support. Doctors were able to take better care of Eva, now that they had a diagnosis.

Benefits of Genetic Testing

Genetic diagnoses are made through diagnostic testing or genetic testing (or both) by a medical geneticist or other genetics expert. A genetic diagnosis may:

- Help doctors take better care of a child with special needs.
- Provide a name for a condition and answers for a family.
- Guide a family to community resources geared to a specific genetic condition.
- Connect a family to other parents of children with the same diagnosis.
- Provide the opportunity to join clinical trials on a specific syndrome.

Parents of children with genetic syndromes often become experts on that syndrome; some parent social media groups have led to the development of foundations for research.

Limitations of Genetic Testing

- Genetic testing does not always provide a diagnosis or an answer.
- There are different types of diagnostic genetic tests. Working with a genetics expert ensures that the right test is ordered and administered.
- There may be no direct benefit to a child, even if a genetic diagnosis is made.

Risks Associated with Genetic Testing

There is a risk for genetic discrimination with all genetic tests. Currently, there is a federal law in place called GINA (Genetic Information Nondiscrimination Act), which prevents health insurers, group health plans and employers from discriminating on the basis of genetic test results. However, this protection does not extend to long-term care, life or disability insurance. It also does not apply to individuals who are in the military. To find out more about GINA, visit **www.genome.gov/10002328/genetic-discrimination-fact-sheet.**

Genetic testing may reveal information that can be unexpected or upsetting, such as a family member having a genetic disorder or biologic relationships not being as stated.

Genetic counseling is recommended before and after genetic testing. For more information, visit:

Rady Children's Institute for Genomic Medicine
www.radygenomics.org

National Society of Genetic Counselors (NSGC)
www.nsgc.org/findageneticcounselor

American College of Medical Genetics (ACMG)
www.acmg.net

A Note about Direct-to-Consumer (DTC) Testing

Direct-to-consumer genetic tests (DTC), such as 23andMe, allow consumers to "order" genetic testing without guidance of a medical professional. These are not diagnostic tests, so results from them should not guide medical decision-making. If something runs in your family, see your doctor, who may provide a referral to a genetic specialist for appropriate risk assessment and diagnostic testing. If you decide to send in a sample, read information provided by the company carefully and ask these questions:

- Do I know what this company does with my genetic information? Will they share it with third parties?
- Will the company protect my genetic privacy? How do they secure the information collected from me?
- Who will help me interpret the results: genetic tests do not always provide a "yes" or "no" answer for risk of developing a genetic disease.
- What do the results really mean for my health? Do not make decisions based on incomplete, inaccurate or misunderstood information.

Seek a Professional

Consultation with a medical geneticist or genetic counselor is recommended if you're considering genetic testing, so that risks, benefits and limitations can be thoroughly reviewed. A genetic professional will record a thorough family history to assess whether or not an individual is at increased risk for a genetic condition. The goal is to ensure that people who choose to have genetic testing do so with informed consent and the correct genetic test is ordered. In some cases, a parent or individual may decline to have genetic testing done due to concerns about genetic discrimination and privacy or the potential for psychological harm. ❖

Lisa Salz, MS, LCGC is a genetic counselor at Rady Children's Institute for Genomic Medicine.

BE FABULOUS!
Donate to the
Special Needs Resource Foundation of San Diego.
Providing resources for parents of children with special needs!

DO IT TODAY!
Donate at
SNRFSD.org