

Why Should We See a Genetic Counselor?

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At birth, Samantha and Derek's first child seemed small but healthy. As she grew, the couple discovered Sydney had learning and medical problems. The couple has had a hard time caring for their child's special needs, but they have done the best they can under the circumstances with the support of family and friends.

Samantha and Derek yearn for another child. Yet they are concerned the next child will have problems too. The idea of caring for two children with disabilities is more than they think they could bear. What should they do?

Sometimes couples refrain from having another child once they have had one with a serious medical condition. In many cases, however, a genetics professional can reduce a couple's anxiety by discovering the recurrence risk in their family is actually much lower than the couple first believed.

In Samantha and Derek's case, a pediatric geneticist diagnoses their child, Sydney, with Williams syndrome. Williams syndrome typically causes mild to moderate mental retardation and heart defects. Fortunately Sydney's heart problem is relatively minor and does not require surgery. Individuals with Williams syndrome often have outgoing personalities and sometimes have extraordinary hearing and musical abilities. Interestingly, the severity of symptoms of Williams syndrome can differ greatly among affected individuals.

Once they learn of their new pregnancy, Samantha and Derek are referred to a genetic counselor. During their consult, the couple learns that Williams syndrome recurs in some families. Individuals with Williams syndrome are missing a part of chromosome 7. This phenomenon is known as a chromosome deletion.

The good news is that testing is available to look at the gene(s) on chromosome 7 responsible for Williams syndrome and tell whether either Samantha or Derek has a deletion on that chromosome. Sometimes the affects of having William syndrome are mild enough that a parent is only diagnosed once their child is diagnosed because of their more dramatic symptoms.

The couple's blood is drawn following their meeting with the genetic counselor. A few days later the counselor calls. Although neither of them have obvious features of Williams syndrome, Samantha and Derek are very nervous since they are already pregnant. The news is good: neither parent has a deletion. This makes it very unlikely that their new baby also will have Williams syndrome.

The couple later chooses to have an amniocentesis, and happily a normal Williams syndrome test result is reported by their genetic counselor for their baby. Months later, another baby girl is born. Sydney is very interested in playing her toy piano for her new

sister, Emily. Samantha and Derek are happy to have another child; this one smiling like her sister and also healthy.



GeneCare Medical Genetics Center, in Chapel Hill, offers genetic consultation and testing, as well as prenatal ultrasound services. Couples who have a family history, are at least 35 years of age, or are concerned about the possibility of having a baby with medical problems should seek a genetic counselor in their area. For more information please call (919) 942-0021 or 1-800-277-4363 or visit www.genecare.com