Illumina Patient Case Study: Allison Querni

Lessening the Burden of Not Knowing

Allison Querni, pregnant with twins, used the verifi® Prenatal Test to get fast and accurate results after an ultrasound exam raised some concerns.

Introduction

Allison Querni, 34, was pregnant for the first time with twins. After getting married two years ago, Allison and her husband decided to start a family. Following a number of setbacks, they decided to undergo in vitro fertilization (IVF) to increase their chances. The IVF was successful, but Allison and her husband still had some anxiety due to the difficulties they experienced getting pregnant.

Twelfth-Week Concerns

The anxiety intensified at Allison's twelfth-week ultrasound, which showed a possible concern with one of the babies (“Baby B”). Ultrasound findings indicated that Baby B’s nuchal translucency (the space between skin and tissue at the back of the fetus’ neck) was in the upper limits of the normal range. In some cases, this increased measurement indicates a chromosomal abnormality, such as Down syndrome. Algorithmic calculations conducted using Allison’s blood screens also showed an increased risk for Down syndrome. With these factors, Allison was referred to a high-risk specialist, Dr. Martin Chavez, Chief of Maternal Fetal Medicine at Winthrop-University Hospital.

Non-invasive Prenatal Test (NIPT)

Dr. Chavez reviewed Allison’s case and decided that more details about the abnormality could be obtained using the non-invasive verifi Prenatal Test. The verifi Test involves a simple blood draw from the mother to analyze fetal chromosomes. Allison was relieved, as she was not comfortable with the added risks of a genetic diagnostic test via chorionic villus sampling (CVS) or amniocentesis. She desired the reassurance of knowing about her baby’s health, but did not want to risk having a miscarriage due to an invasive procedure.

In addition to reduced risk, Allison received results from the verifi Test within one week. Upon receiving the results, which indicated there was no aneuploidy detected, her anxiety turned to relief. Allison’s stress level decreased and she felt better about her pregnancy. Knowing that Baby B did not have a chromosome abnormality, Allison felt a sense of calm and believed that she could tackle any other obstacles her pregnancy presented.

“All even though you still feel scared for the baby, a blood test really lessens the burden.”
Looking Forward

Allison’s experience with the verifi Test gave her a positive feeling about NIPTs. If she has any future pregnancies, she would use the test again. The lack of side effects, quick turnaround time for results, and non-invasive nature of the test made that decision a “no brainer” for Allison.

In April 2014, Allison gave birth to two healthy babies.

Learn more about the verifi Prenatal Test at www.illumina.com/verifipatient

Summary

Overview

Allison Querni was pregnant with twins when an ultrasound raised concerned about the chromosomal health of one of the babies.

Challenge

Allison wanted the reassurance a prenatal test could provide, but was not comfortable with the risks associated with invasive tests.

Solution

Her doctor recommended the non-invasive verifi Prenatal Test.

Benefits

Allison received the conclusive results of an invasive test without the risk, reducing the stress of her twin pregnancy.