#RareDiseaseDay Information Kit

Let’s honor this #RarestDay by spreading the word

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Carrier Screen

Myriad genetics

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Throughout this guide, you will find useful information to build awareness of Rare Disease Day and the benefits of carrier screening in the community.

can make undiagnosed rare disease #MoreRare

Leap Day comes every 4 years—and this year, February 29 is Rare Disease Day, too. But did you know? Most kids living with a rare disease must wait even longer to get a correct diagnosis, specialized care, and support.

4.8 years

is the typical waiting time for a rare disease diagnosis[1]

The good news? Many rare diseases could be diagnosed sooner, if every family received the genetic insights of a carrier screen during or prior to pregnancy.

Spread the word throughout February—especially with friends and family who may start a family in the next few years. Together, we can make a difference before the next #RarestDay. No family should have to wait 4 years to find help.

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Ashley's inspiring story

Screening made all the difference

Ashley's family has experienced pregnancy with and without genetic carrier screening. Read their story to better understand why medical experts recommend that every family consider this screen before or during pregnancy.

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Ashley is 26, **pregnant**, and experiencing a **complication-free pregnancy.**

Ashley delivers Christopher.

At around 18 months, Christopher has **not yet started walking.**

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Ashley and her husband learn about the Foresight Carrier Screen, get tested, and find they are **both carriers of spinal muscular atrophy (SMA).**

Christopher gets **medical interventions and extensive physical therapy,** and eventually **learns to walk with braces.**

Ashley is **pregnant again** with Emily. Because they know they are SMA carriers and there is a 25 percent chance Emily could be affected, **they have an early intervention plan.**

Emily is delivered and is **positive for SMA**. The **early therapeutic interventions** mean that Emily hits her milestones and shows **no symptoms of SMA.**

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RARE DISEASE FACTS

Rare disease isn’t rare at all

By definition, individual rare diseases affect 200,000 people or less. But the overall community living with rare disease across the globe is anything but small.

There are **10,000+**

types of rare and genetic diseases[1]

1 in 13

live with a rare disease in the U.S.—likely including people you know[2]

400 million

people experience rare disease, globally[1]

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RARE DISEASE FACTS

The journey to diagnosis can be isolating

Families navigating mysterious symptoms and healthcare choices may not know where to turn for help. Until they have a correct diagnosis, it may be hard to identify the right dedicated support groups for them, with a community of families who have all been there, too.

7 providers

are often consulted to get a correct diagnosis[1]

2,237

rare disease organizations support patients in the U.S.[3]

Up to 80%

of rare disease is genetic.[1] The risk can be detected before or during pregnancy with carrier screening.

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HOW FORESIGHT HELPS

Early insights matter

If every family received a genetic **Foresight Carrier Screen** before or during pregnancy, many could find answers, specialized options, and rare disease support faster. About 4 out of 5 rare diseases has a genetic component.[1]

With the genetic insights Foresight provides, families can start addressing the risk of a rare disease before the baby is delivered—or even before conception, when screening is used in family planning.

OPTIONS THAT MAY HELP BEFORE PREGNANCY

In vitro fertilization: embryo genetic testing can be done before implantation

Family-building options: from adoption to sperm or egg donation, and beyond

OPTIONS THAT MAY HELP DURING PREGNANCY

Plan for diagnostic testing

Consult experts

Choose a specialized delivery facility

Find support groups

Explore treatment options

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HOW FORESIGHT HELPS

Foresight is for every parent-to-be

Anyone can have a baby with an inherited genetic condition, no matter our family history or ancestry. Many of us are carriers and don't even know it. And only Foresight is reliable for patients of *every* ancestry.

1 in 300

pregnancies are affected by the conditions Foresight screens[4-6]

88%

of people who carry a risk of cystic fibrosis, spinal muscular atrophy, and fragile X syndrome have no known family history[7]

Only Foresight is >99%

reliable for patients of all ancestries, across the vast majority of screened conditions[8]

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Social Media Posts

Share these with friends and family, especially anyone who might start a family in the next few years.
Together, we can make a difference before the next **#RarestDay**.

Suggested copy: You and your partner share many things, but did you know that you might also share a risk of passing on a genetic condition to your child? The Foresight® Carrier Screen can help you identify whether you both carry mutations in the same gene that could affect your baby with a serious condition. Knowing this risk early on gives families multiple ways to plan a family or prepare for a child with a rare disease. Visit www.Myriad.com/Rare to learn more. #Foresight #CarrierScreening #PrenatalScreening #RarestDay #RareDiseaseAwareness

Suggested copy: Rare diseases are not as rare as you might think. There are 30 million people in the United States living with a rare disease, and 80% of those rare diseases have genetic origins. Most babies with genetic conditions are born to parents with no known family history of that disease, like the Pantiers. If you are planning a family or are already expecting, you can help make undiagnosed rare diseases #MoreRare by taking the Foresight® Carrier Screen with your partner. Visit www.Myriad.com/Rare to learn more. #Foresight #CarrierScreening #PrenatalScreening #RarestDay #RareDiseaseAwareness

Suggested copy: Carrier screens can help couples create a roadmap to plan their families and pregnancies. Understanding the risk of passing on an inherited condition to your baby is the first step.
Visit www.Myriad.com/Rare to learn more. #Foresight #CarrierScreening #PrenatalScreening #RarestDay #RareDiseaseAwareness

Suggested copy: The American College of Obstetricians and Gynecologists recommends that all pregnant patients get offered a #CarrierScreen to learn whether they or their partners could be carriers of a genetic condition that they could pass on to their child. Sometimes parents choose not to take the test because they “think they would not do anything differently” if the test showed they were both carriers. In fact, there are many actions parents can take to plan and prepare their family to care for a child who may have a rare disease. Visit www.Myriad.com/Rare to learn more. #Foresight #CarrierScreening #PrenatalScreening #RarestDay #RareDiseaseAwareness

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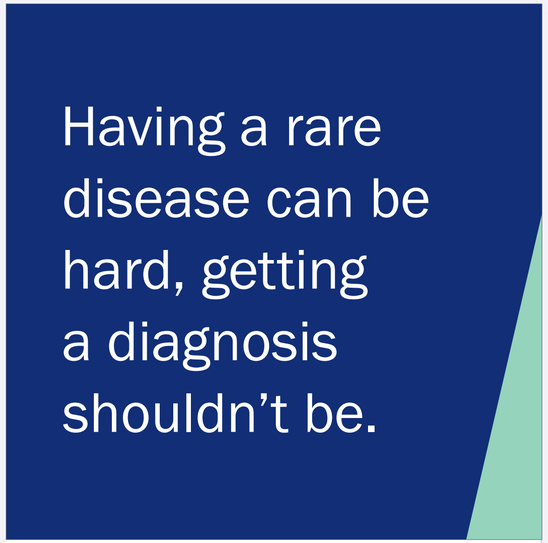
Suggested copy: Considered the #RarestDay on the calendar, February 29th comes only once every four years. Most children living with a rare disease must wait even longer than four years from the onset of their symptoms to finally arriving at a diagnosis. But many rare conditions could be diagnosed sooner if parents benefitted from the insights of a #CarrierScreen prior to or during pregnancy. To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening #RareDiseaseAwareness

Suggested copy: For some rare diseases, time is of the essence. Without diagnosis and treatment, a child's life expectancy is deeply affected. Thus, the earlier the diagnosis the better. This #RareDiseaseDay and every day, Myriad Genetics is dedicated to providing patients and providers with tools like the Foresight Carrier Screen. To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening #RareDiseaseAwareness

Suggested copy: "Through Foresight® Carrier Screen, my husband and I learned that we were carriers of rare diseases never before noted in our families. Though we were at a low risk of having a child with one of these diseases, we were able to educate our extended family on their potential risks." To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening #RareDiseaseAwareness



Suggested copy: Foresight® Carrier Screen can help identify couples who are at risk of passing on a serious inherited genetic condition to their child. This can help reduce the time until diagnosis and provide prospective parents more options to plan and prepare. To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening #RareDiseaseAwareness

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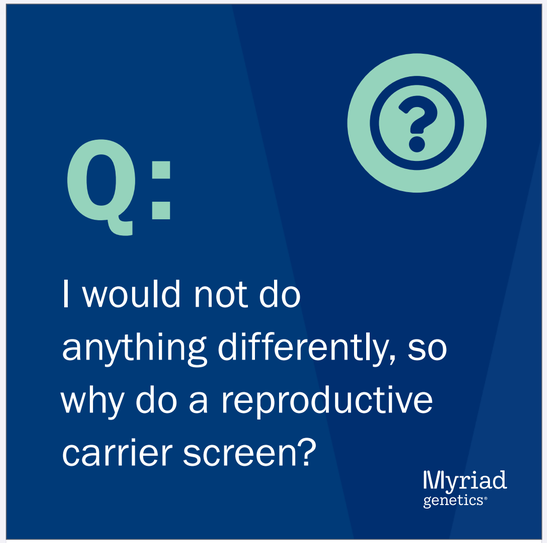

Suggested copy: A: In fact, there are many proactive things you can do when armed with the knowledge a carrier screen may bring. For example, expectant parents who find out they may have a child affected with a rare disease can join support communities, arrange for delivery at a properly equipped facility, and connect pre-partum with pediatric specialists, to name a few. To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening #RareDiseaseAwareness

Suggested copy: Many of us are carriers of inherited conditions and don't know it. In fact, 88% of individuals who are carriers for cystic fibrosis, spinal muscular atrophy, and fragile X syndrome have no known family history. Foresight® Carrier Screen can screen your genes for serious, prevalent, clinically-actionable inherited genetic conditions. This #RareDiseaseDay, give yourself and your partner the gift of knowledge carrier screening can bring as you plan your family. To learn more, visit www.Myriad.com/Rare. #Foresight #CarrierScreening #PrenatalScreening
#RareDiseaseAwareness
Reference: Archibald AD, et al. 2018.

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