

MORINFORMATION

INSIGHT INTO CHILDREN'S GLAUCOMA AND CATARACTS

This publication is dedicated to the memory of Dr. Donald Morin, an internationally acclaimed expert on pediatric glaucoma and cataracts



*Helping children see
the sun,
the moon,
and the stars*



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Family Story: Marissa Terrell

It was the start of our 10th year of marriage and my husband and I were going to be a family of three! We were enjoying every moment of our perfect pregnancy and preparing for our first baby to arrive. I had just turned 37 and I remember the doctor asking me about testing. My response was, “No worries! If we find that our baby girl needs treatment, we will do our homework, get the right specialists and it will be okay.”

On March 23rd, 2009, I got up, got dressed and went to work. I was sitting at my desk when my water broke. Then, 20 hours of labor and a c-section later, Marissa arrived! The first thing I remember was everyone’s facial expressions while I waited on the other side of the curtain to see my daughter. In a conversation with my mother and husband, the doctor said, “No, the redness won’t go away. We will talk more tomorrow.”

What did that mean? I wanted to see Marissa even more to figure out what they meant. When I looked at her and saw the port wine birthmark and her swollen eye I thought, “She can’t go to school like this. We are going to need help.” I knew our world was going to be different than other new parents, I just didn’t know yet how different.

The next morning at the crack of dawn, our hospital room became a revolving door. Doctor after doctor arrived. Every doctor that came by left me with the feeling that more was coming, and I needed to prepare. When a dermatologist arrived, I thought, “Wow! They’re taking Marissa’s ‘birthmark’ pretty seriously to send a specialist so soon!” Finally, the pediatrician explained what could, should, or might happen with Marissa... starting with a CAT scan. I was screaming on the inside. “What does all this mean? Yesterday, they said it was



just a birthmark and bruising, now my day-old baby needs a CAT scan!”

The results came back. They had been looking for calcifications (*whatever that meant!*) and luckily didn’t find any. The neurologist explained that she may have [Sturge-Weber Syndrome](#). My head was spinning. I had no idea what that was, but we focused on the good news and started to relax and get ready to take Marissa home.

When we came home we had our first visit with the pediatric glaucoma specialists from Wills Eye Hospital. At the point we met with Dr. Alex Levin. He explained that we need to see her under anesthesia to see what was happening and to get the pressures. Fortunately, her pressures were fine and no glaucoma.

As the weeks turned into months, Marissa had check-up after check-up. I started to learn all I could about Sturge-Weber Syndrome.

I came across [The Sturge-Weber Foundation](#) and we decided to attend a conference as a family. We met Karen Ball for the first time. She scooped up Marissa like we were old friends, adored her as if there were no birthmark and she had nothing but love for our Marissa. This was the first person that didn’t look at Marissa with any judgment. I never felt so comfortable or free to talk about the syndrome Marissa might have and what we were going through as a family. We’d found a new home.

Thank goodness we had them because when Marissa reached 10-months our world turned upside down. Marissa cried and moaned all morning, then out of nowhere she vomited, and back-to-back seizures began. It took nearly five days to control the seizures. It was

official. We had a Sturge-Weber Syndrome diagnosis and we were sent home with medication.

It happened again when she was two and a half years old. These seizures were even harder to control. We were sent home again after five days with even more medications. This even became even more frightening. It was time for her next exam under anesthesia and Dr. Levin came out to talk to us. He explained that she had glaucoma and we began down the road of eye drops. The exam otherwise was unchanged and we hoped that the drops would work. That day we realized everything they said could happen with Sturge-Weber was happening.



Marissa has had some challenging appointments and we have been through some different drops and oral medicines to control the glaucoma. She tolerates the medicines well. We do have to check every three months to make sure we are on track. Dr. Levin and Wills Eye have always taken good care of Marissa at every turn.

Today, Marissa is 11 years old and is holding steady. Her seizures and glaucoma are controlled. She had many laser treatments for her birthmark too. The newest symptom of the syndrome are auras and possible migraines. She may have more doctors than the average 11-year-old and have a harder time doing the things her friends can do, but her life doesn't stop for Sturge-Weber Syndrome. She's a champion for positivity and hope and she teaches me something new every day.

We always try to keep Marissa protected. We slow down when she's sick to reduce the risk of seizures and glaucoma. With Sturge-Weber Syndrome Marissa has accomplished so much in her 11 years. She's gone on vacations, advocated on the hill, participated in dance, sports, art, Girl Scouts and music, and she swims every week. Yes, she will struggle physically because of her syndrome, but she's not afraid to tell her story and believe in herself! We don't know what tomorrow will bring for Marissa, but we're so grateful that we have the blessing of front row seats watching the rest of her story unfold. To learn more about Sturge-Weber Syndrome and hear more stories

about wonderful people like Marissa, please visit sturge-weber.org



Encouraging Age Appropriate Self Advocacy

Julie McIntyre, Mom of 2 with Congenital Glaucoma

No one is prepared to parent a child with Glaucoma or Cataracts. Understanding how to properly teach our children to self advocate as well as be an advocate for

their child is important. We must evolve with our children as they grow and change themselves. We need to conduct ourselves in a way that both nurtures and empowers our children in this lifelong journey that we will face together. One way is to Talk openly at home and

with family about your child's diagnosis. This can be very beneficial as everyone is learning and growing together.

Self advocacy starts early. Many children will not feel totally comfortable telling an adult that something isn't working for them especially if they are in a group of their peers. It is our job as parents and caregivers to encourage our kids to be an active participant in learning how to talk about what they are experiencing. Using age appropriate content, you child should be encouraged to share every day what went well and what didn't go so well. By having these active conversations frequently, we will then begin to understand the nuances of our specific child's needs and help develop language your child can use on their own. Every kid with a visual impairment will not fit into the same need category!

Throughout this entire process, in addition to us , as parents, being able to accurately describe what works best for our children, our children will also begin to learn how to articulate what accommodations or modifications they may need in a classroom or other social/ extra-curricular settings. We can then begin to work with them on techniques to approach these sometimes uncomfortable conversations. Our children will need to overcome these obstacles throughout their lives and the sooner we teach these skills, the better we are equipping them for their future.

As a younger student your child will not be able to give a detailed description of their diagnosis to his or her teacher, but they will know if their eyes feel "funny" or if their light sensitivity is too much today. Practicing how to describe experiences or role playing different scenarios will be helpful in self advocating. See below for more suggestions.

As your child gets older and moves through school, they will find what does and does not work for them. While it will be important for your child to communicate, keeping the lines of communication open between yourself and the teacher is absolutely critical in your child's success. During the first week of each school year, make it a point to contact the teacher(s) to discuss your student's needs. Requesting an IEP for your child so that they are provided with the necessary accommodations is helpful

for everyone involved. Most educators are not familiar with low vision accommodations and IEP meetings are not always at the very beginning of the year. Reaching out and explaining your child's diagnosis and needs will help ease any anxieties you and your child may experience. Be aware that you can request an IEP meeting to discuss concerns or changes in accommodations at any time, not just when it is "due"

The following are examples of age appropriate methods of self-advocacy for your child to practice at home, community, and school.

Toddlers to Pre-K

The light is bothering me

I can't see the pictures

Kindergarten- Grade 3

I can't sit here, the light makes it too difficult to see

I need to stand at the back of the line walking up or down the steps

I need to wear my hat and sunglasses when I go outside

Grade 4-6

For my materials in class, I need to use a larger, bolder font

I need a buddy when I am going to an unfamiliar area

Grades 7-12

At this point, your student should be an active member of their IEP process. If they are not invited to the meeting you can request they be there. They can give you valuable input on what needs to be done to make them a more successful student. Let their voices be heard at the table!

By allowing our children to add their personal opinions in their everyday school atmosphere, we are empowering them to self-advocate appropriately and respectfully for life.

***The view in this article are the author's own and do not represent the opinion of the PGCFA.*

PGCFA Award for Best Research in Glaucoma 2020: Rate of Complete Catheterization of Schlemm's Canal and Trabeculotomy Success in Primary and Secondary Childhood Glaucomas

American Journal of Ophthalmology, Year 2020, Volume 212, Pages 69-78

Carin Rojas and Brenda L. Bohnsack
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Glaucoma affecting children often requires surgery to lower eye pressure and preserve vision. Trabeculotomy is sometimes used for childhood glaucoma that works by opening the "drain" of the eye, called the trabecular meshwork, to allow excess fluid to leave the eye more easily. Trabeculotomy surgery is performed from a small incision on the "white part of the eye" (sclera). From this incision, the "drain" is first identified and then opened with specially designed instruments. Traditionally, trabeculotomy surgery, using an instrument called the Harms Trabeculotome, is able to open approximately 1/3 to 1/2 of the drain from the single incision. More recently, a flexible lighted tube has been used in order to open the entire drain (360 degrees) from the one incision. A handful of studies have shown that this procedure may give longer control of eye pressure in some children. However, the instrument is often difficult to pass through the entire drain, especially in children with congenital eye diseases.

In this study, we assessed the ability to pass the tube through the entire drain in different types of childhood glaucoma. We found that the tube was only able to be passed through the entire drain in just over 2/3 of eyes with juvenile open angle glaucoma, and only 1/3 of eyes with other types of childhood glaucoma such as primary congenital glaucoma and glaucoma following cataract surgery. To date, eye pressure continues to be controlled in all remaining eyes in which the instrument was able to open up the entire drain. This is in contrast to eyes in which the tube was not able to be passed through the entire drain. In eyes in which the Harms Trabeculotome were used, eye pressure has been controlled in approximately 2/3 of eyes.

The use of this new instrument in trabeculotomy surgery can be effective in obtaining eye pressure control in some children. The ability to pass the instrument through the entire drain is challenging with best chance of doing so in eyes with juvenile open angle glaucoma (JOAG), but is much more limited in other forms of childhood glaucoma. Although this study did not compare the procedure to other option for glaucoma surgery, it does suggest that there may be a role for this procedure as yet another option for childhood glaucoma.

PGCFA Award for Best Research in Cataract 2020: Does the Aphakic Eye Contribute to the Visual Field With Both Eyes Open?

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There is a belief that doing cataract removal on infants and children with congenital cataract in only one eye is not beneficial if the straight ahead vision in that eye may always be less than the other normal eye. This is largely due to failure of the brain to develop the vision in that eye: a condition called amblyopia (sometimes referred to as "lazy eye"). Even with patching of the good eye to strengthen the bad eye, the vision may never fully recover. The purpose of our research was to see whether there are benefits to performing this surgery, even in children where the vision in the eye that has the cataract surgery remains poor after surgery.

In this study, we performed visual field testing on nine patients between 10 and 26 years old, who previously had congenital cataracts in only one eye and had cataract removal in infancy. Visual field testing is a way to measure side vision. Our visual fields were taken with both eyes open, and also just the right and left eye, on all patients.

We found that the field of vision with both eyes open increased significantly compared to the good eye alone, indicating that operating on the poorer seeing eye does add some benefit even if the straight ahead vision cannot be improved by much.

Visual function is not only a measure of straight ahead vision, but also visual fields (side vision), color vision and contrast sensitivity. Our research suggests that removing the cataract when only one eye is affected is beneficial in improving the visual field with both eyes open, even in cases where the straight ahead vision in the eye that had surgery is poor and may not have improved significantly.

It may be useful to measure patient improvement with visual field testing rather than only straight ahead vision testing.

Areaux RG Jr, et al: Trabeculotomy Ab interno with the Trab360 Device for Childhood Glaucomas.

American Journal of Ophthalmology. January 2020; volume 209: pages 178-186

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To lower the high eye pressure in eyes with glaucoma, doctors can use surgery or medications. Eye surgery for glaucoma tries to find a way to lower eye pressure in two ways: (1) improving the natural drain of fluid leaving the eye, or (2) creating a new drain for fluid leaving the eye. In children with glaucoma in infancy, we often try method #1. We can either do this by opening the drain from the inside of the eye (called "ab interno") or from the outside (as externo). Many surgeons prefer ab interno surgeries, because the second type damages the tissues around the eye that might be needed later for further surgery. The gold standard ab interno surgery is called a goniotomy. A goniotomy is when a needle or tiny knife is passed through the clear front part of the eye that then cuts open the natural drain directly in front of the colored part of the eye. Using this method less than half (120 degrees) of the drain can be easily opened at a time. The authors in this article use a different ab interno surgery tool called the Trab360, which is no longer available, but it is very close to a similar tool available today called the OMNI. The Trab360 approach is similar



to goniotomy, but, on entering the natural drain of the eye, it releases a thread into the drain allowing the entire drain (360 degrees) to be opened in one surgery. The authors are surgeons from 4 different hospitals in Minnesota, California, and Florida. They included patients of varying ages who had the Trab360 surgery and were followed after surgery for at least 3 months. The authors looked at eye pressure and the number of glaucoma medications before and after surgery. Surgery was called successful if: (1) the eye pressure was lowered to 24; (2) no extra surgery was needed after to lower the eye pressure; and (3) there were no severe complications.

Forty-six eyes were included in the study. The average patient age was approximately 1 year with a range of 1 month to 27 years old. More than half of the patients were younger than 2 years old. Six eyes had a previous operation. Patients were usually seen after surgery for more than one year. Before surgery, the average eye pressure measured around 30. After surgery the eye pressure was on average about 20. The average number of glaucoma medications needed before surgery was 2.6, and after surgery was 1.6. Success was found in 67.4% of patients. Success was even higher (70%) in patients who had Trab360 as their first surgery. Success was also higher in patients with primary congenital glaucoma (83%), which is a childhood glaucoma thought to be due to a bad drain rather than any other cause or disease. Fifteen eyes were considered failures, of which 13 needed further surgery. Two eyes had a problem during the surgery where the Trab360 tool went too far

into the drainage system, causing too much drainage, lowering the eye pressure too much. Both of these eyes needed further surgery to fix this problem. A common but minor problem was blood build up in the eye during and after the surgery.

The authors feel that the Trab360 (and they feel perhaps the OMNI, which wasn't tested in this study) is a safe surgery for children with glaucoma. The surgery was especially a success in children with primary congenital glaucoma, where the drain is thought to be bad in the first place.

This is similar to results for goniotomy. However, goniotomy is much cheaper, causes less damage inside the eye, and is more available as it does not require the special Trab360 tool, takes less time, and may be easier to perform. If the outcomes are the same, maybe the extra time and risk of opening the drain for 360 degrees is not justified since the smaller amount of opening in goniotomy seems to have the same result. It is very difficult on this relatively small sample from a wide variety of glaucoma types and patient ages to conclude that the Trab360 (or OMNI) is safe, but it does offer surgeons another option in treating childhood glaucoma in certain circumstances. Perhaps future studies could

Jackson CM, et al: Unplanned Returns to the Operating Room within Three Months of Pediatric Cataract-Related Intraocular Surgery: Indications and Risk Factors

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Cataract in childhood usually needs surgery. When a child has any kind of surgery, every parent worries about possible complications. But how common are such complications and what do they look like? Four authors from Storm Eye Institute in Charleston, South Carolina reviewed medical records of children who had cataract surgery at their center from 1991-2018. They tried to determine the frequency and type of complications that occurred within 90 days after the initial surgery and needed another surgery. The initial cataract surgery technique was done according to age:

look more carefully at particular kinds of glaucoma, at more specific ages and compare the currently available OMNI to goniotomy.

some children had glasses or contact lenses after surgery and others got a standard plastic intraocular lens (IOL) to replace the natural lens. Children who had other pre-existing eye abnormalities, in addition to the cataract, were excluded.

Out of 1,392 cataract operations, only 3.3% (46/1392) required a second operation for a complication and 0.14% (2/1392) needed two operations within 90 days after cataract surgery. The complications that needed surgery included removal of pieces of the initial lens that were left behind or regrew a bit, glaucoma (increased pressure inside the eye), scar tissue that was blocking vision or distorting the structures inside the eye, abnormal tissue positioning inside the eye and, in one case, a tiny piece of fiber left accidentally inside the eye. Complications were more often seen in children under 1 year old at the time of the cataract operation and cataracts at any age that were caused by an injury. Without these two risk factors, the complication rate was even lower with only 1.1% of children needing a re-operation.

The findings of this study are very similar to what other researchers at other centers have found: complications of cataract surgery are very uncommon.

Of course other complications, such as glaucoma, can still occur after 90 days, but at least this study tells us that in the first 90 days of cataract surgery it is very unlikely that a reoperation will be needed, especially if the child is over 1 year old and if the cataract was not due to an injury. This should provide great reassurance to families about the safety of cataract surgery today.

Genetic Counseling: When is it Needed?

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As genetic testing becomes more widely available, it is more important than ever that medical genetics professionals are available to provide genetic counseling. Genetic counseling is the process of helping people understand and adapt to the implications of genetic disease, both medical and psychological. It involves the patient as well as interested family members. Genetic information can be complex. Trained professionals, such as genetic counselors, carry a skill set to simplify and communicate this information in way that is meaningful to patients and their families.

Many forms of pediatric cataract and glaucoma can have non-genetic causes [See *MORINformation Fall 2019*]. In most cases, genetics is a factor. Many families have questions, including, for example: “What caused my child’s eye condition? Can I have another child with cataract or glaucoma? Do we need to worry about other associated health problems in the future? Genetic counselors can assess these risks and the likelihood of a genetic cause, and provide information to address these

questions. In some cases genetic testing may provide further answers, and in others, testing may have less benefit. When applicable, genetic counselors provide guidance for appropriate test selection. This is critical because ordering the wrong test, or testing unnecessary genes, may cause more harm than good; it could provide information an individual or their family did not want to know, yield confusing information that does not address the family’s initial questions, or miss the target completely and potentially give families misinformation. The genetic counseling process also facilitates informed consent. Every genetic test carries different risks, limitations, and potential benefit to the patient and their family, about which they should be well informed before they choose to have testing. This ensures a person and/or their family understand the possible testing outcomes and can manage their own expectations.

If testing is pursued, genetic counseling is equally important as test results are returned to the family so they have assistance in understanding the meaning of a test result. Through counseling they learn how it might change the child’s medical management, if at all, and what implications there may be for other family

members. The genetic counselor can provide support as they process this information. Genetic counselors also facilitate communication and interpretation of results to other members of a child's care team in order to optimize medical management.

Genetic testing must include pre- and post-test genetic counseling. Genetic counsellors can be relied upon to help you consider whether you want to pursue genetic testing. And if you decide against genetic testing, the genetic counsellor is still there for you should you have questions about a possible genetic link, implications for your family, the risk of having another affected child and whether other testing for associated conditions is needed.

In fact, genetic counseling by a trained genetics professional is highly recommended whenever genetics are considered regarding your child's body or eye disorder.

You can find genetic counselors in your area at the following sources:

National Society of Genetic Counselors (US and Canada):
<https://www.nsgc.org/page/find-a-genetic-counselor>

American Board of Genetic Counseling (worldwide):
<https://www.abgc.net/about-genetic-counseling/find-a-certified-counselor/>

Meet Our Scientific Advisory Board: Dr. Ken K. Nischal

Dr. Nischal is Chief of the Division of Pediatric Ophthalmology and Strabismus at Children's Hospital of Pittsburgh of UPMC, Director of Pediatric Program Development at the UPMC Eye Center, and Professor of Ophthalmology at the University of Pittsburgh School of Medicine.

Dr. Nischal attended King's College Hospital Medical School of the University of London and completed ophthalmology residencies at both Birmingham and Midlands and the Oxford Eye Hospital in the United Kingdom. He completed his pediatric ophthalmology fellowship at The Hospital for Sick Children in Toronto. Prior to joining Children's Hospital of Pittsburgh in 2011, Ken was at Great Ormond Street Hospital for Children in London, UK.

Dr. Nischal is one of the pioneers of pediatric corneal transplants. Children from all over the world come to see

Dr. Nischal because of his expertise in helping children with genetic corneal diseases. His research focus is on evidence based protocol led clinical care with clinical outcome measures to drive clinical care. His main areas of clinical research are anterior segment developmental anomalies affecting the cornea, lens and trabecular meshwork. He has published widely on the topics of pediatric cataract, glaucoma and cornea as well as craniofacial anomalies.



He is a member of the Scientific Advisory Board of the Pediatric Glaucoma and Cataract Family Association (PGCFA) and has contributed articles and responses to our Ask the Doctor feature. He is the founder and co-director of the World Society of Pediatric Ophthalmology and Strabismus, an outstanding organization with members from around the globe.

The Robison D. Harley, MD Childhood Glaucoma Research Network (CGRN) International Pediatric Glaucoma Registry

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CGRN Newsletter 6/01/2018

The Robison D. Harley, MD Childhood Glaucoma Research Network (CGRN) International Pediatric Glaucoma Registry

Childhood glaucoma is a rare disease. Research on rare diseases has always had a hurdle: small study sizes. Since this project was introduced to the PGCFA in the Spring 2016 MORINformation newsletter, international clinicians and researchers have been coming together to tackle this challenge through the **Robison D. Harley, MD Childhood Glaucoma Research Network (CGRN) International Pediatric Glaucoma Registry**. This registry acts as a centralized database that leverages contributions from centers across the world to create larger and more powerful studies. Any physician who cares for children with glaucoma is encouraged to join the effort.

Since 2016, there have been a number of exciting updates to the Registry. A second phase of development has added the ability to add data regarding the outcomes of patients: how they responded to different medicines and surgical



procedures as well as vision outcomes. This addition will allow researchers to better understand how childhood glaucoma is treated in varying parts of the world and the impact of particular treatments. The Registry infrastructure has also been developed to allow interested participants to deposit genetic material from children with glaucoma, into a DNA bank that is linked to the Registry data. As advancements towards "personalized" medicine progresses, understanding what we have learned from the registry data in the context of a patient's DNA or learning about genes that cause childhood glaucoma will become even more impactful. A genetic information company, Invitae, which maintains a number of registries in its Patient Insights Network™ program, has taken over the Registry maintenance. The Registry Advisory Board has also been expanded to include representation from Invitae and from international representation (from Australia).

After a moratorium on data entry to allow for final development and new maintenance by Invitae, the Registry's secure online portal is now live! Including those that are participating and those that are in the process of joining the registry, at the time of this writing the Registry represents a collaborative effort from doctors in Argentina, Australia, Canada, Chile, China, Ecuador, Germany, India, Iran, Israel, Italy, Japan, Jordan, Korea, Nigeria, Saudi Arabia, South Africa, Spain, Taiwan, Thailand, the United Kingdom, the United States, and Venezuela. As the de-identified data from several hundred patients are already included in the database, a number which is growing every day, the first research inquiries are being proposed. It is certainly an exciting time for pediatric glaucoma around the world!

There is no charge to join, participate, or develop research from the Registry. Its development, maintenance, and growth has come about through the generosity of the Robison D. Harley, M.D. Fund of the Wills Eye Alumni Society. Although the Registry is hosted at Wills Eye Hospital, the Registry belongs to the global community of all those that care for children with glaucoma.



The PGCFA Needs Your Help!

*Together, we can help children
see the sun, the moon and the stars*

Don't forget that if you make a charitable contribution to the PGCFA you will receive a tax receipt. Not only are you helping children and their families, you receive a personal benefit as well.

Your contribution will ensure the PGCFA can continue to-

- Provide you with important information on treatments, new and innovative research and stories you can relate to through our website and newsletter
- Host our annual Education Day
- Encourage new discoveries through our research awards

The Pediatric Glaucoma and Cataract Family Association is a volunteer group, committed to promoting the quality of life for children with glaucoma and cataracts and their families by:

- Providing information and education
- Providing support

Your Support Will Make a Difference!

For more information, visit our website at www.pgcfa.org To support the Pediatric Glaucoma and Cataract Family Association, please send in your donation to our address as listed below. Please be sure to include your mailing address so that we may send you a charitable tax receipt. Or, to make an online donation, please visit our website at www.pgcfa.org

Here are three ways you can make a difference:

1. Direct your annual United Way pledge to the PGCFA by listing the PGCFA's charitable number on the back of your pledge form. Our charitable number is: 31954
2. Make a one-time or recurring donation online at www.pgcfa.org it's fast, easy and secure. All major credit cards are accepted.
3. Mail your contribution to:

PGCFA C/O Michelle Breslin
4411 Woods Edge Ct.,
Chantilly, VA 20151

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We sincerely thank you for your kind support in the past and we look forward to your continued support in helping children see the sun, the moon and the stars.

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