



The Journey Through Diagnosis



A guide for families of children with a genetic condition





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Introduction

The Purpose of This Guide

The purpose of this guide is to provide information and support to families whose child has been identified with a genetic condition. This guide is written from the family perspective and includes information they felt was important when facing a new diagnosis. The material includes advice and tips to help “guide” you on your journey.

Who Created This Guide?

Multiple families who have a child (or children) with a genetic condition wrote this guide. Some of those families learned of their child’s diagnosis prenatally, some through newborn screening, while others waited months or even years. Their children are affected by a wide variety of conditions including spinal muscular atrophy, cystic fibrosis, PMM2 congenital disorder of glycosylation, sickle cell disease, Pelizaeus-Merzbacher disease (PMD), MECP2 duplication syndrome, metachromatic leukodystrophy, Rett syndrome, phenylketonuria (PKU), Down syndrome, trisomy 18, chromosome 2p deletion, DYRK1A syndrome, and others. You will hear their voices in the following pages. They share what they have learned and want to pass on to others from their experience.

How to Use This Guide

This guide is intended to empower families. It may not answer all your questions, but it can get you started. There is no right or wrong way to use the information. You can read cover to cover, or just the parts of interest. There’s a lot of information, and some of it may not apply to you in the moment. You can come back and revisit chapters as needed. The families who created this guide hope you find it useful, making the “journey through diagnosis” a little easier.

Sincerely,

The Family Forum

Region 4 Midwest Genetics Collaborative



Region 4 Midwest Genetics Collaborative

Region 4 Midwest Genetic Collaborative (Region 4) is a collaborative network of families, genetic service providers, newborn screening laboratories, and leaders in public health genetics from Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin. Region 4 Midwest uses a regional approach to improve access to services, quality care, and genetics expertise in a medical home environment that is culturally sensitive.



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Chapter One

Emotions



If you are reading this, your child or someone you know has been diagnosed with a genetic condition. Life may be taking a different path than you imagined. Some children are diagnosed prenatally or through newborn screening. Other children are identified later, after delays or concerns appear. Not every genetic condition has a name.

Regardless of circumstances, you may be feeling a range of emotions. Not every person feels the same thing. There is no right or wrong way to feel during this time.

Emotions may change from day to day, or even hour to hour. Depression, anger, fear, and uncertainty are just a few things you may feel. These are strong emotions, making it difficult in the beginning. Each person's reaction is unique. It's not unusual to feel like you are on an emotional rollercoaster at first.

One thing to remember is a diagnosis doesn't change who your child is. It is important to still celebrate all that is in their life. Over time you will figure out how to play and have fun, and also remember to laugh. You will move forward and find joy again.

How you think you'll feel...



How you may really feel...



***"My child is not his diagnosis. The words don't define him.
He is first and foremost a child."***

Feelings of Grief

You have experienced a loss if your child's condition impacts, limits, or changes your dreams for them. It is okay to allow yourself time to grieve. The way each person experiences grief is unique and can be scary and overwhelming. There are typically thought to be five stages of grief. You may go through all or some the stages. You may experience each stage in a different order, more than once, or go back and forth. The stages of grief are denial, anger, bargaining, depression, and acceptance. You may not recognize what you are feeling is part of grief or understand why you're feeling this way. Friends and family may not recognize your grief, which can intensify feelings of being alone.

Symptoms of the different stages can vary. For example, denial may include feelings of disbelief or numbness. Anger is often the result of feeling powerless and may cause difficulty with close relationships. Depression can include feelings of sadness, hopelessness, or irritability. Grieving is a process. No one can predict exactly how long it will take. Sometimes a crisis or particular event can cause you to experience the stages all over again.

Knowing that your reactions are common may help. You will need to find new ways to manage these feelings. Recognition of what you are feeling and why can be a first step. If any of the emotions become too much, it is okay to seek out professional help. Suggested ways to find support are identified later in this chapter, but your doctor, a trusted friend, or clergy member may be a good place to start. Acceptance is about coming to terms with a new reality. It doesn't mean you like it, but you find a way to manage.

Feelings of Guilt

You may also experience feelings of guilt. You may wonder if you could have prevented your child's condition. You may feel responsible. You may question if you didn't know your family history. Some people may experience guilty feelings about what was done, or not done, during pregnancy. Others may feel frustrated because they can't "fix" what is wrong. These are common concerns and feelings.

As you move through initial diagnosis to managing your child's condition, new challenges may set off new feelings. Having a child with medical challenges can be overwhelming, and the need to understand why can be a strong feeling. Gaining knowledge about your child's condition may reduce the stress associated with feelings of guilt.



There are doctors and counselors who specialize in genetics. Genetics is the study of how traits such as hair color, height, or risk for disease are passed from parents to their children.

A Geneticist or Genetic Counselor can tell you more about certain genetic conditions and how they occur. They can help you understand family history and impact on other family members. They may also help you identify what treatments or specialists work best with certain genetic conditions.

"It's overwhelming to focus too much on what may or may not happen in the future...Remember to find the joy in your life."

Coping Techniques

There are many different ways to deal with the emotions you feel after your child's diagnosis. The following are parent recommendations. What works for one person may not work for another. It is important to identify ways to manage your emotions and stress in ways that are healthy for you.

- Educate yourself as much as possible. Find reliable information about your child's condition. This will help you to ask better questions and seek the best care for your child. (See Chapter 3, Getting Help & Information)
- Find support. Family and friends may be a source of comfort and healing. Make an effort to keep in touch with people, even if it's hard at times. Seek out new sources of support. Connecting with other families who have similar circumstances might help.
- You may find that you enjoy and take comfort in activities you do by yourself. Running or other exercise, gardening, fishing, meditating, crafts, reading, and journaling are all examples of healthy outlets.
- Take care of yourself. Pay attention to your physical, mental, and spiritual well-being. Keep your medical and dental appointments, especially for preventive care. Make an effort to establish routines that are familiar and comforting for yourself and others in the home. Make time for yourself to relax and continue to do activities you find enjoyable. This can be especially difficult, but it can help you avoid a crisis later.
- It is okay to ask for and accept help that is offered. You may feel that you have to do everything and it is your responsibility. You do not have to do it alone. Make a list of specific things friends, neighbors, and family can do that you can share when help is offered. For example, they might help with cooking a meal or running an errand for you.
- Remember it's a marathon, not a sprint. Life may have changed drastically for your family. You may feel a sense of urgency to do everything at once. You need to pace yourself. Prioritize what is most important in your life. Slow down and take a deep breath once in a while.



The Diagnostic Odyssey—When Your Child's Condition Remains Undiagnosed

Sometimes a child has symptoms or delays that do not match known conditions. Your child's condition may be genetic in nature but not yet diagnosable through current medical technology. For some, it may take years to get a diagnosis, if at all. You may feel isolated. You may feel frustrated and confused by the doctors not being able to diagnose your child. It may be hard when well-meaning friends and family ask questions. You and your family are not alone. There are still resources and support available to you. You will learn how to navigate in this area too. Having to accept the unknown can be challenging, but you can find a way to live with it.

Chapter Two

Family Relationships



Getting a new diagnosis doesn't just impact you and your child, it can add stress to the entire family. Siblings, grandparents, and other extended family may feel the effect. Your child's medical condition is a unique experience your family will share. It can change how your family functions, both positively and negatively. There may be tough medical decisions, financial concerns, and time constraints, all of which may alter family dynamics.

How your family is impacted depends on many things, such as your child's condition and severity, family coping skills, financial means and other resources. A child with complex medical needs may put extra demands on the family system, often for an extended period of time. As with any other major life event, your family will learn to adjust and adapt over time.

***“There is a hidden beauty in where you are today,
so start looking and find someone who will
hold your hand along the way.”***

Parents or Caregivers

Sometimes a new diagnosis can cause conflict or stress in your marriage or other significant relationship. You and your partner may handle things differently. It is okay to let your partner deal with feelings in his or her own way. Your partner is often the closest person to your situation and may end up feeling the brunt of your anger and pain. Remember to be kind and find an outlet for your emotions.

Give your relationship time to deal with the added stress. Use available care resources to schedule some time together as a couple. This can be challenging when time is precious.

Now, more than ever, it is important to communicate. Find ways to keep your partner up to date if the other can't attend an appointment. Think of yourself as a team and try to help one another. View this as an opportunity to grow.

As with your own well-being, it is okay to dedicate time and energy to your relationship. Seek out professional services if needed. Despite challenges, it is possible for your relationship with your significant other to thrive. Some couples find it can actually foster a deeper relationship. Sharing this experience can help you grow as a couple.



Siblings

Having a child with a genetic condition may also impact siblings. Regardless of age, children pick up on stress or activity around them. They may have questions that are difficult to answer. Be honest and answer their questions the best you can and keep your answers at your child's age level.

Just as you may experience a range of emotions, so can siblings. It may swing back and forth between positive and negative emotions. Some children will want to help with caregiving tasks, while others may be less helpful or even resentful. It may change over time. The following are suggestions from other families.

- Remember they are still children
- Help identify and model healthy ways to express their feelings
- Seek out opportunities to involve them in typical childhood experiences
- Spend one-on-one time with them when possible
- Give ways to answer questions other people may ask them

Despite the challenges, many adults raised with a sibling with a genetic condition say their experience was primarily positive. They say it helped them acquire patience, perseverance, and compassion. They learned to view people with different abilities as a natural part of life. As they grow, siblings may have questions about their own health or the health of their potential children. Help them to find good resources to answer their questions as they get older.



Extended Family

Grandparents can be affected as well. They can experience their own emotions, as well as feel their child's pain.

They also need good information and support. Educate and share resources with them that are helpful for you.

Your extended family may have questions. It is up to you how much you include family in your inner circle. It is okay to set boundaries. Some people find that extended family is a source of much needed help, while other family members may mean well, but aren't a source of support at all.

Extended family may want to help but aren't sure how. Share what would be most helpful. Give them specific, practical ways they can assist, such as helping with childcare, cooking a meal, or running errands. This will allow them to contribute.



Chapter Three

Getting Information & Support

Educating yourself and getting services and supports will help you feel more in control of your situation. With the Internet, families have more information than ever at their disposal. Not all sources provide accurate information, however. There are many resources in your community, your region, your state, and online to help your family.

Getting Information

There are a wide variety of places to get information and help, depending on where you live. Below are a few places you can reach out to for help and information.

- **Primary or Specialty Care Providers**—Ask for a care manager or coordinator if one is available. Their job is to help identify resources and support within your state and local community. A care manager or coordinator can help with more than just medical issues related to raising a child with complex medical needs. Helping to connect you with resources is also part of their job.
- **Children’s Hospitals**—Besides medical care, hospitals may offer family support groups, social workers, and more. Some hospitals have family centers for support, information and resources.
- **Early Intervention**—These programs are sometimes referred to as “Birth to Three.” Every state offers some type of program to babies and toddlers if they have delays or specific health conditions. An evaluation will be done to determine if your child is eligible. There are a variety of different services they may cover.
- **Community Mental Health**—These centers offer community-based services for individuals with mental health disorders and developmental delays. Community-based care means services and supports are delivered within the community you live. This is more effective and helps families achieve better outcomes.
- **Parent-to-Parent**—These organizations provide information and emotional support to families of children who have special needs. They match parents with an experienced, trained parent for support. Visit www.p2pusa.org.

Parent Tip!

Whenever getting services from an organization or agency, be sure to ask them if there are more places you can go for help and information. You can also ask other parents or caregivers what has been most helpful to them.

“In the beginning I worried about every little thing. There are still worries of course, but we are better able to put them in perspective.”



- **Baby's First Test**—A website that brings together resources and information about newborn screening and baby health for parents and health care professionals. Visit www.babysfirsttest.org.
- **Genetic Alliance**—An organization promoting good health for individuals with genetic conditions. They provide information on health, disease intervention and ways to participate in research through various websites including *Genes For Life* and *Disease InfoSearch*. Visit www.geneticalliance.org.
- **Family to Family Health Information Center**—These federally-funded, family-led organizations provide support, information, education, and training to families of children with special health care needs. There is a Family to Family Health Information Center in each state. Often they work together with Family Voices (see below).

• **Family Voices**—A national network of state-specific organizations that promote quality health care for all children and youth, particularly those with special health care needs. They make sure the family voice is included to improve health care programs and policies. Visit www.familyvoices.org.

- **Disease-specific foundations and organizations**—Look for a large, well-established group that can provide information about your child's condition and support for your family. Some organizations have local, regional or national conferences. These organizations may also offer information on research and clinical trials, if available.

Evaluating Resources

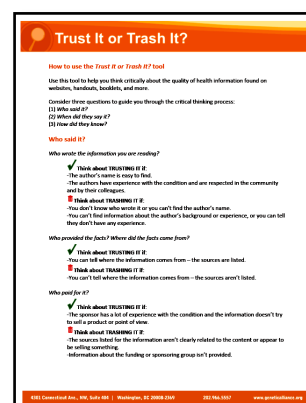
You may find yourself overwhelmed by the constant flow of health information from many sources, both reliable and unreliable. Some of the information is good, and some of it isn't. You need to filter out the irrelevant or inaccurate information so you get good health information that can have a positive impact on your child's life.

One resource available to help you is called "*Trust It or Trash It*" from Genetic Alliance. This tool helps you think critically about the quality of health information. It teaches you to ask yourself three simple questions:

- 1) "Who said it?"
- 2) "When did they say it?"
- 3) "How did they know?"

This gives families a way to review health information, especially from online sources. It is natural to want to believe that information out there can help your child, but it is important not to let that hope cloud your judgement. The tool is available online at:

www.trustortrash.org



Finding Support

There are ways to find support as you go through this journey. There may be people in your area you enjoy spending time with, or you may choose to connect with others online. You may be able to meet parents who have a child with the same diagnosis. Talking to someone who has gone through similar circumstances can be emotionally uplifting.

Usually you get support from the people around you—your informal support system. This might be your friends, neighbors, family, or others you have regular contact with. You may find your relationship with some people changes over time.

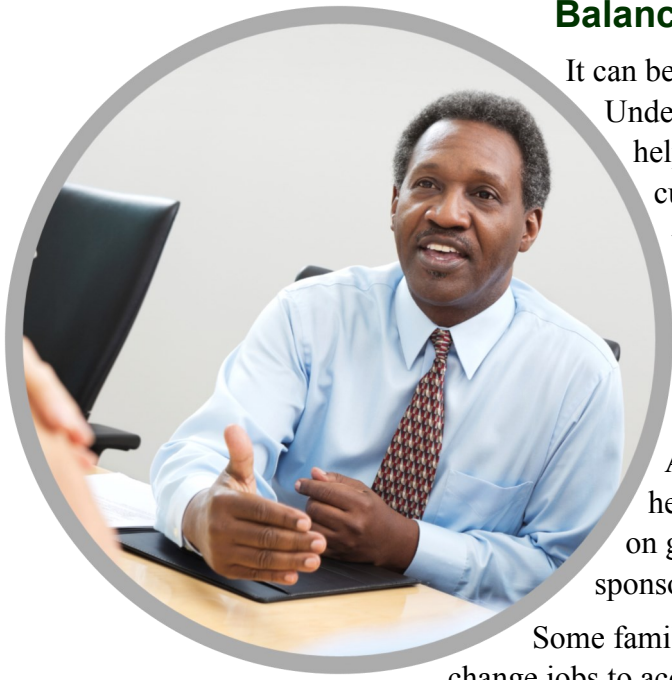
You may become closer to some and grow apart from others. You may need to make new friends. This is not uncommon with any major life event, and all of us change and grow due to our life circumstances. Sometimes the people you interact with in your everyday life can give you the support you need. Other times you have to identify different ways to get support. Here are a few examples of ways to find support.



- **In-person support groups**—Community centers, children’s hospitals, places of worship, and local disease-specific groups are a few of the organizations that may offer in-person support groups. Some groups meet weekly, some monthly, some less frequently. It is alright to try several groups to find one that fits you best.
- **Online support groups**— This may be in the form of an email listserv or a group on Facebook. Online support lets you connect with other parents regardless of where you live. Some families have found this especially helpful when their child has a condition that is less common or rare.
- **Peer-to-Peer support**— A trained support partner or mentor is someone who has experience providing emotional and informational support to another parent. Parents may be matched based on diagnosis or similar circumstances.
- **Spiritual support**—A church, synagogue, mosque, or other places of worship may offer a community of support and comfort. You may find that you have questions about your faith. Talking with your clergy or other spiritual leader may help.
- **Professional support**—Seeing a doctor or professional counselor is another option. You may benefit from talking to someone who is neutral. They are able to offer you new ways to manage your feelings. This is especially helpful if you are struggling with overwhelming or persistent feelings of sadness or depression. If you feel in danger of hurting yourself, contact 911 or the National Suicide Prevention Hotline at 800-273-8255, or go to your local hospital immediately.

“Look for allies through finding connections with other parents and providers. Once you find that connection, trust usually follows, which in turn helps the (care) navigation process.”

Balancing Work and Family



It can be difficult to balance the demands of home and work. Understanding the challenges posed by your situation can help you be proactive. You can explore options with your current employer to determine how to manage this.

You may have concerns that having a child with a chronic health condition may impact your work. This can be especially tough if you are a single-parent. You may worry about reducing the number of hours you work or losing your job, resulting in a loss of income.

Another job-related challenge for caregivers is keeping health insurance coverage. Be sure to review the section on getting coverage on page 15 in case you lose employer-sponsored insurance coverage.

Some families reduce hours, take less demanding positions, or change jobs to accommodate their unique situations. Be sure to ask about different options, such as working from home or a different schedule to allow you to work more effectively. Even if you do not qualify under the Family Medical Leave Act, good communication with your employer may help.

Despite the challenges, many caregivers maintain a career while providing care for their child. Some parents have also found new and rewarding careers that support their parenting roles, or allow them to use their experience as a caregiver of a child with medical challenges. The skills you are developing now can be useful in the job arena. Some examples include time management, budgeting, setting goals, analytical thinking, and relationship building. You are also learning other traits that employers value such as perseverance, flexibility, and determination. Like many of the challenges you may encounter, you will adapt and determine what is best for you and your family.

Family Medical Leave Act (FMLA)

Family Medical Leave is one option to help protect your employment if you require time off to care for a loved one. It allows eligible employees to take unpaid, job-protected leave for specific family and medical reasons. It also allows continuation of group health insurance coverage under the same terms as if you had not taken leave. Be sure to communicate with your employer if this impacts you. Your child's doctor may have to fill out paperwork for you to qualify. More specific information is available on the US Department of Labor's website.

www.dol.gov/whd/fmla/

“My priorities, my time and my own goals have had to shift. I’ve not had to abandon these things; however, I do look at each of them within the context of the responsibilities of being a parent and caregiver to my child. “

Chapter Four



Medical Care

Your child may require a lot of medical care. You may not have prepared for this before your child's birth. It can be hard to work with different doctors. It is important for you to have a voice in your child's care. You will become an expert on your child. Don't be afraid to ask questions and speak up when you need to. You are an important part of your child's care team. You want providers who recognize your role. One way to do this is to have a medical home.

What is a Medical Home?

Having a medical home may benefit your child. A medical home is not a specific place or building, it is a way of delivering health care. The American Academy of Pediatrics (AAP) describes it as a model of care that is accessible, continuous, comprehensive, family-centered, coordinated, compassionate and

culturally effective. This means you have one central place to go that meets the needs of your child and family. Your child's medical home may be with a primary care doctor or it may be with another health care provider.

What to Expect from a Medical Home

Your child's health care provider should see you as an important part of your child's care. You will want to be able to get to the office easily, and to reach someone quickly when needed. It is reasonable to expect the doctor and staff treat you with respect, even if your family does things differently from them. It is important to have issues explained to you in a way you understand, and in your language if needed. It also means that your child's doctor is willing to work with other doctors and hospitals. They will coordinate care so your child doesn't have unnecessary tests or delays in getting needed care. A medical home will be with you every step of the way and help when you need it.

Benefits of a Medical Home

Here are some of the benefits for your child and family if your provider is a medical home.

- Your child sees the same doctors and office staff regularly. They get to know your child and family.
- The doctor partners with you in your child's care. This means the doctor is honest and respectful and you learn from each other.
- Your family gets help finding support and resources to meet your child's needs and challenges.
- They work with you to support good health and quality of life as your child gets older.

Care Coordination

Care coordination is one benefit of having a medical home. Region 4 Midwest Genetics Collaborative defines care coordination as a needs driven, team-based process that values families as essential partners. It is intended to integrate care among multiple service providers, enhance the caregiving ability of the family, and maximize the potential of the child.

It means that there is someone at the doctor's office who helps you put all the pieces together. This may be a social worker, a nurse, or a care manager. Even when you have someone to help you, your role is important. You are the constant in your child's life. You may not always have a choice of doctors, but you can make an effort to have an active role in your child's care.



“Don’t be afraid to search and switch specialists until you find the ones you love and who are invested in your child.”

Finding Providers Who are “All-In”

It is good to have medical providers who work with your child and family as a team. One parent described these providers as being “all-in”. This means they will respect your family's beliefs and decisions, and be a trusted partner and resource. There are many different ways to find this type of provider, including the following.

- Ask for recommendations from parent groups or disease-specific organizations. Other families can be a valuable resource. They have experiences they can share with you. Keep in mind, however, that everyone's experiences can differ. Look for a good fit for your family.
- Connect with a social worker who works in health care. There are hospital-based pediatric social workers or case managers who can help you find providers who can meet your family's needs.
- Research medical journal articles related to your child's condition, and make note of the authors and what hospitals they are from. These doctors are more likely to have treated patients that are similar, participated in research, etc.

Think of your first visit with a new provider as an interview. You are interviewing for an important position in your child's life. It is okay to ask questions. In fact, this is vital! First and foremost, you should feel comfortable. If you are comfortable, most likely your child will be too. Look for signs that the doctor is open to learning and sharing. You want a provider who will actively listen to you and your concerns. They should be supportive and talk to you—not at you.

Good relationships take time and work, but your child's care will benefit from these efforts. More importantly, you are modeling good health partnerships for your child.



Hospital Stays

You may find yourself in the hospital with your child. This may be a planned visit or it may be unexpected. While it isn't always possible to plan ahead, here are some parent suggestions that may help you if your child is hospitalized.

- Keep a “to-go” bag ready for hospital stays. It should be something you can literally grab as you leave your house. Include a change of clothes for you and your child, toiletries, chargers for electronics, snacks, change for vending machines, and a comfort item (such as a toy, stuffed animal, video, etc.) Label personal items with your child's name.
- Create a care notebook with your child's vital medical records and information. It can be a physical binder or electronic

version. Include a list of current medications and dosage schedule, any allergies, and history of medical procedures. Share your child's likes and dislikes. There are online care notebook templates if you need ideas to get started.

- Know where you can find a list of your insurance's preferred providers for medical supplies or durable medical equipment. Depending on the reason for your child's hospital stay, you may need to get special supplies or equipment before going home.
- Have all providers explain to you who they are, what they are doing and why they are doing it. If you don't understand, ask again. Have them explain until you are comfortable you understand. This keeps you in the loop and informed about your child's care.
- Look into lodging information in case you need it. Some hospitals have a Ronald McDonald house, however you usually cannot reserve a room ahead of time. Some hotels offer special hospital rates. The hospital may have a list of these hotels, or you can ask other parents or a hospital social worker for suggestions.
- Have a plan for the care of other children and pets, especially if unexpected stays are common. Create a list of people available with their phone numbers and keep it in your “to go” hospital bag. Have a few back ups in case your primary person isn't available.
- Be aware of typical bills that might come due while staying at the hospital, such as mortgage, rent or utilities. You might want to set up automatic payments if possible so it's not a concern.

Parent Tip!

Your child still needs regular checkups for things like hearing, vision and dental care. You may need providers who are familiar with your child's genetic condition. Sometimes a condition can impact other areas of health as well, causing secondary ailments to arise. For example, some children only eat pureed food. This can cause teeth to grow crooked or be slow to break through the gums. They may also have increased plaque build up because they can't chew food. In this case, having a dentist who knows about and can treat special conditions in addition to providing regular checkups is helpful.



Medical Formula and Food

Some medical conditions require your child to use special formula and food. The formula and food may be needed to keep your child healthy. This impacts not only your child, but your entire family.

With most children, you introduce new foods without a lot of direction. With a child on a medical diet, you will work closely with a dietician and specialty clinic staff. They can help you learn how to find nutrition that best suits your child's health condition.

You will become a master of reading food labels. You will learn to look for ingredients that need to be avoided. You may even shed a happy tear when you find food you can purchase at the local grocery store instead of having to order online.

A big challenge can be when you want to go out to eat. You must plan ahead when going to a restaurant or events to make sure there is something your child can eat. You will learn where the best places are for your entire family to eat, including when you will need to pack special food and formula.

Learning from other families with similar diagnoses may be helpful. They can answer questions about daily challenges, such as:

- How do you go to a fast food restaurant and order?
- What types of fruit snacks are ok for your child?
- How do you find a daycare that will adhere to your child's diet?
- Where do you order special food and how do you get medical formula?

Things that seem simple to other families may feel overwhelming when your child has a special diet to follow. Each step is a transition. It is a time to learn and grow. You are your child's teacher. This is important so he or she will be able to manage independently one day. Use your resources to help you and your child adapt to what may be a lifetime of following a special diet.

"While there are still bumps along the road, these are not the huge hurdles of years ago."

Insurance Coverage for Medical Formula and Food

Insurance coverage for special food and formula varies from state to state and you might hit difficulties.

There are insurance coaches available to assist with navigating the insurance maze. You can find an insurance coach through established support groups or through a specialty clinic.

An insurance coach has worked with other families like yours. They can help with special insurance coding for coverage of formula and food.

"No" doesn't always mean "No" and you will learn how to advocate to get what is needed to keep your child healthy.

Additional Care Resources

Sometimes you need to consider additional medical care choices for your child. It is helpful to learn about the different types of care that may be recommended.

Palliative Care

Palliative care (pronounced pal-lee-uh-tiv) is specialized care for patients with serious health conditions. It focuses on providing relief from symptoms and stress of ongoing medical treatment.

Palliative care involves clinicians from different areas, such as medicine, nursing, social work, child life, spiritual care and different kinds of therapies. Palliative care strives to provide comfort for you and your child. It improves quality of life: physically, psychologically, socially, emotionally, and spiritually.

There are no time restrictions. Palliative care can be received by patients at any age, any time, at any stage of illness, whether or not it is life-limiting. It can be used to avoid making hasty decisions during a time of crisis or acute illness. Most insurance plans, including Medicaid, cover palliative care.

Hospice Care

Hospice care is designed to support the patient and family during the end-of-life process. Hospice care focuses on quality of life and comfort rather than aggressive medical treatment. The goal is to give the patient comfortable, pain-free days. It allows your child to live each day as fully as possible. End-of-life care is difficult to discuss. It is okay to think about what you want for your child before it becomes a concern. When hospice is needed, the doctor will make a referral. The hospice team will meet with the patient and family, usually within 48 hours. The team writes a care plan that includes your input. Hospice can be covered in a variety of settings, and is typically covered in part by health insurance.

Respite Care

Respite care is a planned break from caregiving. It can be helpful for families who provide round-the-clock or long term, constant care of a child with ongoing health issues. Respite care can relieve the ongoing stress of providing care. Parents may hesitate to seek or use respite care. It can be hard to leave your child in the care of someone else. However, there is evidence that respite care benefits both parents and child.

Respite services can be formal or informal. Informal respite may be having a family member or friend care for your child to allow you time to do something other than caregiving. More formal programs may include both in-home or out-of-home care for a specific time. You may have to ask or advocate for funding for respite.



Parent Tip!

The learning curve is often steep when your child is first diagnosed. You are usually trying to learn a lot of new information at the same time you are caring for your child (or children!)

Be kind to yourself and others. You can't (and won't) learn it all at once. Don't waste precious energy on beating yourself up if you make a mistake. Expect some growing pains.

Chapter Five

Medical Insurance



You may find that having a child with a genetic condition will suddenly require you to become an “insurance expert.” This can be difficult. Insurance coverage is complex. Navigating insurance is one of the main challenges parents face.

In order to avoid problems, it is helpful to understand how your coverage works. Get a copy of your benefit plan’s certificate and riders and read them. Whenever you get a bill, compare it with records from your insurance. If it doesn’t add up, ask questions. Understand your rights and how to appeal when you don’t agree with a decision.

Find people who can help you. Ask what new terms mean when you hear them. It is like learning a new language. With practice, you will learn how to navigate through insurance issues.

Getting Help with Insurance

You may find that you need help with insurance issues. There are a number of places you can go to for help. It may vary depending on what type of insurance your child has. Here are a few of the people you may ask for assistance with insurance issues:

- Customer Service Representative
- Case Manager
- Medical Provider’s office staff
- Insurance Advocate
- Human Resources Representative
- Pediatric Social Workers
- Insurance Navigators
- State Commission on Insurance

Parent Tip!

Keep a record of anyone you speak with about your insurance benefits or claims. Record the name of the person you talked to, the date, time and number you called in a notebook.

This can be very helpful if there is a problem later. Sometimes you have to go up the chain of command by speaking with a supervisor. Knowing who told you what is very effective as you advocate for your child.

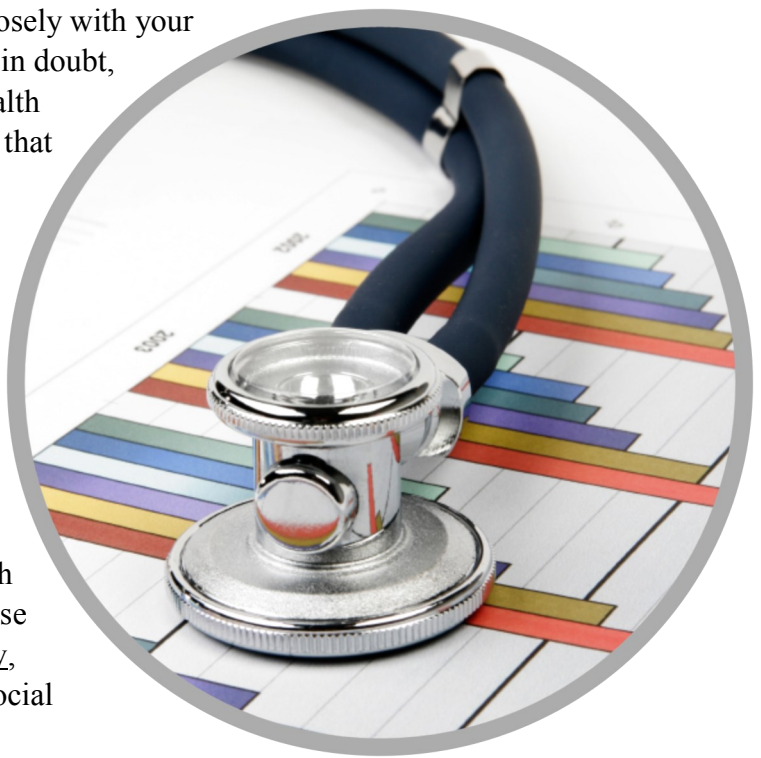
Getting Additional Coverage

Having enough health care coverage may be another challenge. Some families find that having a child with complex medical care means a lot of medical bills. You may want to re-evaluate your insurance plan at the next opportunity to make sure it is still the best one for your family’s needs. Find someone who can help you understand your coverage needs if necessary. There may be programs in your state that help cover medical costs, such as Medicaid and the Children’s Health Insurance Program (CHIP). Medicaid offers health coverage to lower income families and sometimes children with certain medical conditions. CHIP provides low-cost health coverage to children in families that earn too much for Medicaid.

Your state may offer CHIP coverage and work closely with your state's Medicaid program. Eligibility can vary. If in doubt, apply anyway. Be sure to explain your child's health coverage needs and ask if there are any programs that can help.

The Social Security Administration also offers some benefits to children with disabilities. The Supplemental Security Income (SSI) program makes monthly payments to individuals who are disabled. Your child, if under 18, may qualify if he or she has a medical condition that meets their definition of disability. Families must fall within certain income and resource guidelines.

In some states Medicaid comes automatically with SSI eligibility. You can get information about these programs at their website www.socialsecurity.gov, calling 800-772-1213 or by visiting your local Social Security Administration office.



Medicaid Waivers (what are they and how do we get one?)

Waivers are one option available to states to allow them to provide medical care services in home and community based settings under the Medicaid program. States can offer a variety of services under these programs. A Medicaid waiver program can be helpful to provide additional coverage.

The waiver program allows states to be more flexible in the use of their federal Medicaid funds. They may cover items not typically covered by Medicaid, or extend coverage to families that wouldn't otherwise qualify. The waiver may exclude your income to allow your child to receive Medicaid coverage. Some states have the Katie Beckett waiver, but there are other names and types of waivers as well. Below are examples of some services and supports that may be covered with a waiver:

- Van lifts
- Home modifications
- Respite care
- Home or personal care services
- Recreational equipment
- Communication or technology aids

This is not an all inclusive list, and benefits vary from state to state. Access to these programs also varies. Contact your county's social or human services department and ask about the waiver program. If the person you speak to isn't familiar with the term, explain your situation. Ask about funding for your child's services and supports. Other individuals who may help include early intervention staff, community mental health, hospital social workers or your child's physician. Be sure to ask about other funding sources besides the waiver.

Just because your child has a genetic diagnosis, it does not mean he or she will automatically qualify. Each child will be assessed to determine if they are eligible. It is important to apply as soon as possible because there may be a waiting list for these programs.

Chapter Six

Finding Your Voice

You may be frustrated by some of the challenges your child and family face. You may think certain actions or policies don't have your child's best interest in mind. Because of this, you may want to speak out and find ways to change what doesn't seem right to you.

Advocacy comes in many forms. It can range from teaching someone about your child to working with lawmakers to get laws passed. Each of us is different in how we advocate and what we choose to advocate for.

Learning to Advocate

The term advocacy doesn't mean the same thing to everyone. One definition is "the act of speaking on the behalf of, or in support of, another person, place, or thing." When you advocate, you are the voice for your child.

Not everyone recognizes families as equal partners. Learning and practicing advocacy skills can enhance your role and confidence in making decisions that impact your child's life. This is especially important when it comes to your child's medical care.

Advocating for your child demonstrates that he or she matters. You model the importance of being involved. Often your child has better health outcomes when you take an active, equal role in care. It can be hard to feel like an equal partner in your child's care in the beginning. Being an advocate doesn't mean you disregard medical expertise and knowledge, it means you recognize and use the unique expertise and knowledge each person brings.

Learning to advocate happens over time. As you advocate for your child, you connect with others who have similar challenges. Often this helps because there is strength in numbers. By working together, you accomplish more than you would by yourself.

Telling and Educating Others

It is up to you what you share with others. Some families feel that sharing their child's diagnosis and challenges is a way to get support, raise awareness or fundraise. They see it as an opportunity to teach others about something which may be new to them. Raising awareness may help your family feel better, and help others understand the challenges and issues your family is facing, or you may choose to only share with a few people close to you. Some people may be on a need-to-know basis. Whichever you are comfortable with is okay. It is important to discuss this with family and friends so everyone is on the same page, and knows what your family is comfortable with sharing with others.



***“My daughter’s love and strength inspired me
to keep going forward.”***

Looking Ahead...

Take a moment now, to look ahead. The future may not be clear, but over time you will develop a vision. There is a lot of information in this guide, what you do with it is up to you.

Many families have shared that they feel isolated at the time of diagnosis. Looking ahead may be difficult. You may feel fear or uncertainty. It can make moving forward hard. It takes time to adapt. Acceptance is a process. It is a process as individual and unique as every one of us.

It may mean accepting life as it is. There are some things you can change and some things you cannot. You have to learn what choices you have, because even though you may be angry and feel powerless, you do have choices.

You have a choice to surround yourself with people who help you. You can figure out ways to develop new traditions, and navigate the journey together, as a family. All families have the potential for resilience. You can maximize that potential by using your experiences to find meaning in everyday life.

Embrace who your child is. Help them become the best version of who they are – regardless of what that is, or how long they are here. Your child will have an impact on you and others. Your child matters. Love and enjoy your child. You may find that along the way, you become the best version of yourself. Do the best you can today, so that you can look ahead to the future with hope.



***“My life took a different path,
but I am grateful now for the
journey I have experienced.***

***I have grown more as a
person. Life as you know it or
thought it would be may have
changed, but change is not
always bad.”***

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