

LIVING WITHOUT A DIAGNOSIS

Road map and navigation
tools for families



INTRODUCTION

Is this booklet for me?

This booklet is written for parents and caregivers of a child with health and/or learning concerns and a suspected genetic health condition or syndrome, but no set diagnosis. Living without a diagnosis can leave you with questions about the future. **You are not alone.** This booklet has advice and information from other parents and families who are also living without a diagnosis.

“My goal is to prevent another family having to go through what we went through to fight for our kid.”

The purpose of this guide is to give a starting point for parents and caregivers who are on the journey to find a diagnosis for their child. Each family’s journey is unique. The experiences of other families can provide you with comfort and insight, helping you to regain control and maximize the potential for your child.

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LIVING WITHOUT A DIAGNOSIS

The Emotional Roller Coaster

COMMON FEELINGS EXPERIENCED BY FAMILIES:

↓ Shock, Frustration, Anger, Fear,
Loss of Control, Sadness, Grief,
Guilt, Worry, Loneliness and
Social Isolation

↑ Acceptance, Courage,
Determination, Hope,
Resilience, Happiness

Living without a diagnosis *is* living with uncertainty

- What does the future hold?
- Not knowing is the worst part. If I knew, I could get on and deal with it.
- If we had a diagnosis, maybe I could find other parents like me, and if they had older children, maybe I would have more of a view of what the future could hold?

- I'm scared of not having a diagnosis, but I'm also scared of having a diagnosis and of what that might mean for the future.



I want to have a name for what I have. If they have a diagnosis for me, they could maybe find somebody else that would be the same as me, and I'd have some idea of what things would be like for me in the future."

- What new curve ball will we be thrown next?
- What new health or learning concern is going to be added to my child's ever-growing list of challenges?
- When will we be surprised by the next crisis, like a major surgery or the sudden onset of seizures?
- How do you work through the anger and frustration when nobody seems to have answers?
- After you get through the initial grief, how do you restore balance?

YOUR CHILD'S HEALTHCARE TEAM

Who's who - getting to know your team

Most people have a doctor who acts as their care coordinator (organizer), usually their family doctor, pediatrician or community nurse. This person makes referrals to specialists and can also help you to keep track of tests/information and bring all the pieces together. Finding a doctor to act as the care coordinator for your child is important.

Once a health and/or learning concern is noticed in your child, you will likely talk to your family doctor or pediatrician first. If they don't have the answers, they may refer you to different specialists. Understanding who these specialists are and what they do can be overwhelming and confusing. It's hard to meet a lot of new people, figure out the health care system, and know who to trust.

Who's who - getting to know your team

Physicians (Doctors):

CARDIOLOGIST: heart doctor

DERMATOLOGIST: skin doctor

ENT DOCTOR: ear, nose and throat doctor and surgeon

ENDOCRINOLOGIST: glands and hormones doctor

GASTROENTEROLOGIST: stomach and bowels doctor

GENERAL PRACTITIONER (GP): family doctor or primary care provider

GENERAL SURGEON: doctor that does belly operations

GENETICIST: doctor that diagnoses genetic conditions

INTERVENTIONAL RADIOLOGIST: doctor that inserts central lines and feeding tubes

NEUROLOGIST: brain, muscles and nervous system doctor

NEUROSURGEON: brain, skull and spine surgeon

OPHTHALMOLOGIST: eye doctor and surgeon

ORTHOPEDIC SURGEON: doctor who operates on bones, tendons and joints

PEDIATRICIAN: kid doctor - makes referrals to specialists; may be the care coordinator. You need a referral from a GP to see a pediatrician

PLASTIC SURGEON: doctor who operates on skin and face problems like a cleft lip or fingers stuck together

PSYCHIATRIST: mental health doctor

RHEUMATOLOGIST: doctor for bones, tendons and joints

UROLOGIST/NEPHROLOGIST: doctor for kidneys and bladder; a Urologist also does surgery

Who's who - getting to know your team

Other Health Experts:

AUDIOLOGIST: tests hearing and fits hearing aids

DIETICIAN: helps with food plan and food issues

GENETIC COUNSELLOR: helps explain birth defects, genetic and inherited disorders

INFANT DEVELOPMENT PROGRAM (IDP) CONSULTANT: helps children under 3 years with or at-risk of developmental delay

NURSE: works in a specialty clinic or in the community; may be part of the care coordination team

OCCUPATIONAL THERAPIST: works on skills

PSYCHOLOGIST: helps with learning skills, feelings and behaviours

PHYSIOTHERAPIST: helps muscles and joints work better

SPEECH AND LANGUAGE THERAPIST: helps with speech problems, teaches sign language

SOCIAL WORKER: gives support, tips and referrals to community-based care



Commonly ordered tests and investigations

To help figure out why your child has a health and/or a learning concern, the doctor will order tests and investigations. These tests and investigations may help to find out why your child is affected and may give a diagnosis.

This is a list of some of the more common tests and investigations. Based on the concerns with your child, one or more of these test and investigations may be ordered:

AUDIOGRAM: hearing test

BLOOD AND URINE TESTS: there are thousands of different tests. They all look at blood or pee to find clues or traces of specific illnesses or health problems

CT SCAN (COMPUTERIZED TOMOGRAPHY):
uses x-rays to give detailed pictures of brain and body

ECHOCARDIOGRAM ("ECHO"): detailed ultrasound of the heart

ECG OR EKG (ELECTROCARDIOGRAM):
measures the heart rhythm

EEG (ELECTROENCEPHALOGRAM): measures brain rhythms, looks for seizures

FEEDING/SWALLOWING STUDY: uses x-ray images or video of the inside of your child's mouth and throat to help determine what your child can safely eat and drink

|| *You can call the lab ahead of time and find out when's the best time to come in for blood draw so you get the best person. And you can also get numbing cream."*

GASTROSCOPY/ENDOSCOPY/COLONOSCOPY: gives a detailed picture of the inside the tummy and gut

GENETIC TESTS: may look at a single gene, many genes or all genes (called sequencing); may look at all or parts of the chromosomes (called a microarray or karyotype)

MRI (MAGNETIC RESONANCE IMAGING): gives a detailed picture of the brain and other internal organs without the use of x-rays

ULTRASOUND: uses sound waves to give a picture of internal organs

X-RAY: gives a detailed picture of bone

Building positive relationships

A positive relationship with your child's healthcare team will make things easier. ***Like with any relationship, it will take time, patience and effort to grow.*** Sometimes it's hard to stay positive, and that can affect how you interact with the doctors.

“ I'm much pickier about who I take my child to see, and I have a lot more clarity at the beginning of appointments about what the relationship is going to be like.”

Finding healthcare providers that are a good fit for your family's needs is an ongoing process. There will be individuals you like and don't like, ones that you will send cards to at Christmas and ones that you hope never to see again after your first appointment. Most likely there will even be a “break-up” or two! Challenges are a big part of the process. Be patient with yourself and others.



Advice to help with common concerns & frustrations

You and your doctor may disagree about a recommendation regarding your child's care.

Be persistent: keep asking questions until you feel like the healthcare provider has answered your concerns to the best of their ability.
It is OK to ask for a second opinion: it doesn't make you a bossy or demanding parent.

It is hard to be told by an "expert" that you are over-reacting about a certain symptom, or not be taken seriously when you raise a valid concern.

Healthcare providers are the medical experts, but YOU are the expert when it comes to your child.
Maintain respectful communication when you disagree with or feel frustrated by a healthcare provider.

I'm tired of having to repeat my child's complicated history over and over to each new specialist and medical resident.

Remind yourself that every new specialist will see your child from a new perspective, and may find new answers.

Bring your list of questions, notes from research or information gathering you have done, and your list of updates on your child's symptoms and current difficulties to each appointment.

You have a gut feeling about a particular concern but your doctor disagrees with you.

Trust your intuition. If you have a gut feeling about a certain concern with your child but your doctor disagrees with you or does not acknowledge your concerns, it is within your rights as a patient to ask for another doctor's opinion. You can ask to be seen by another doctor in the same clinic, or ask your child's general practitioner or pediatrician for a referral to a different specialist.

Sometimes it feels like a doctor is talking "at" you instead of having a conversation with you.

Approach the relationship as a partnership: each appointment should feel like a two-way conversation. Don't be afraid to ask questions! Ask them to slow down or repeat themselves if you missed something or don't understand.

TIP

Parents need to know that they have the right to ask questions, and if they don't understand the answer, to ask questions again. And to ask questions again, and again, until they understand the answer. And that the fault isn't in their understanding, it's in the explaining.

How do I get help when there is no help?

Ask other parents about their experience with specific healthcare providers.

A good doctor is one who is curious.

Express thanks and gratitude to providers who meet your needs and give great service: a thank you card can have a profound impact!

I don't know who to turn to - no one seems to be taking charge or coordinating everything that is happening.

Seek healthcare providers who are curious and express interest in your needs; if a provider does not feel like a good fit, "breaking-up" and getting a referral to another provider may be necessary.

Know that healthcare providers won't have all of the answers all of the time.

Be clear and assertive about your expectations before arrival and at the beginning of appointments with healthcare providers.

Members of my child's healthcare team do not seem to be speaking to each other.

Request a copy of every consult letter, investigation and test report be sent to you at home, and copied to the family physician and/or pediatrician.

MANAGING MEDICAL APPOINTMENTS

Preparation is key

Keeping track of recommendations, follow-up plans and pending results, and handling your calendar of follow up appointments and new referrals can feel like a full-time, exhausting job.

Unfortunately, the burden of keeping track of the health information is often put on families. You cannot assume that each hospital, clinic, and doctor will have access to your whole file. You will likely be the main keeper of the complete information.

Part of your diagnostic journey is the process of becoming the expert on your child. There is no right or wrong way to approach this, and it may take time and some trial and error to figure out how to best document, track and manage things for your child.

I wish I'd known early on how much more organized to be from the beginning with medical care, records, IEP at school, and so on. I always feel like I'm still catching up."

Organizational tips

- ☑ Make a binder with tabs, separated by specialty, to catalogue your child's medical history, reports, specialist consultation letters, imaging (x-rays, MRI, CT) and lab reports.
- ☑ Staple cards and contact information for each specialist to their consultation letter.
- ☑ When you check-in for your child's appointment, ask that a copy of the report (or consultation letter) be sent to you at your home address. Remind your care provider at the end of the appointment that you have asked for a copy.
- ☑ ***It is your right as a patient to get a copy of all reports and letters.***
- ☑ Follow up as needed, to make sure that you get a copy from each appointment, and to get the lab and imaging reports they ordered.
- ☑ When you get a report, file it under the proper section in your binder.
- ☑ Consider bringing your binder to each appointment with a new specialist, as they may not have access to all of the records.
- ☑ Keep a running list of questions for each healthcare provider/specialist, and bring the list to each appointment.
- ☑ Keep notes on the status of your child's symptoms, highlighting improvements, any loss of skills, or other changes. Separate these symptoms into the right specialty as best as you can.
- ☑ ***Always take notes during your child's appointments.***
- ☑ Bring a family member or friend to each appointment (one more set of ears and one more set of hands to take notes).
- ☑ Set calendar reminders to help track important dates, such as appointment times and the date you were told you would hear back from someone.

YOUR SUPPORT SYSTEM

Relationships with family and friends

We are all nurturing many relationships in our lives: you may be in a partnership or have other children; you may have close or more distant relationships with your extended family; and you may have a small or more broad reaching community of friends. The challenges of your journey to find a diagnosis will take a toll on these relationships.

I wish I had known that for all the friends and relationships that would fade, there would be new and more meaningful ones growing.”

Many parents have expressed the common experience of having family/friends that used to be “there” for them, become distant and pull away. They have come to accept that over time, some people drift away.

Your diagnostic journey will be exhausting, and you only have so much time and energy. Deciding which relationships to nurture, and which to let go of, is a necessary part of the process.

It can often be hard for others to acknowledge your child's differences and needs when you are not able to give them a diagnostic label. Some people may be uncertain and express disbelief if you can't give them a specific name or condition to look up. Just as you may need to work through your own denial, so will your family members and friends.

Some parents have described their family members as being overly hopeful, stating that the child will 'grow out of it'. It can be upsetting and tiring when you do not receive the support and validation you need. The last thing you want to do is spend your time convincing others that something is wrong with your child.

My partner and I weren't equally engaged in the search for a diagnosis for our child. It was very hard."

My circle of friends has really shrunk, and it's a better circle! I've got a lot more clarity about the people that I can really count on. But it was a very hard and unpleasant lesson to learn."

A common challenge with extended family members and friends is that they are not in the same head space as you. It is not that they are unkind or not sympathetic, they just may not understand. Over time, you will come to accept that some people may not be capable of ever "getting"

what you are going through. For others, it may just take time for them to integrate into your new rare world. Even if extended family and friends may not fully understand your day-to-day experience, it is so important to ask for help whenever possible. Help from others enables you to manage your responsibilities more effectively.

Relationships with family and friends

- ☑ Use humour as often as possible
- ☑ Make time for family activities and for having fun
- ☑ Express your needs - be honest
- ☑ When you ask for help, be specific
- ☑ Help ease people into your day-to-day with short visits. Once they become more comfortable with the daily tasks of caring for your child, they will feel more confident in taking care of your child, even if just for a short while, to give you breaks
- ☑ Take into consideration the unique strengths of your friends and family when handing out specific tasks
- ☑ Take a team-approach - talk about how responsibilities will be shared or distributed
- ☑ Be respectful and less judgmental of one another, as each person has their own way of dealing with things
- ☑ Go at your own pace - you don't have to share everything with everyone
- ☑ Learn how to set boundaries - you may have to be more firm with certain people
- ☑ Be prepared for everyone offering you advice and decide how to best respond to it
- ☑ Acknowledge that everyone is reaching understanding at their own pace, and it is unlikely you will always see things the same way

TIP

One parent emailed her family and friends a copy of her application for her child's Disability Tax Credit. While this application did not give a name for her child's condition, it reviewed and outlined her child's medical history and ongoing challenges, helping others better understand her child. For background information on the Disability Tax Credit and other Tax Tips, please refer to the Wiki page on the Rare Disease Foundation website.

Self-care: Relationship with yourself

The most important relationship to nurture is the one with yourself.

You will make endless sacrifices and accommodations as a parent, but you must ***put on your own oxygen mask first!*** This is easier said than done, but in order to best care for your child and family, self-care must become a priority. ***Be kind to yourself, trust yourself, and take care of yourself.*** You can't force yourself to relax, but you can seek out ways to help reduce your stress.

Give yourself permission to take a mental break from the never-ending search for a diagnosis -- whether that break is for an hour, a day, or a week -- it is so helpful to give yourself that permission.

If you let it, it will consume you. Find ways to breathe. Helping yourself = helping your family.

Try to maintain a regular hobby, interest, and/or activity as part of your weekly or monthly routine. Maintaining your own passions during this diagnostic journey is important!

TIP

Counselling services can provide much needed emotional support for individuals and families living without a diagnosis. Please see the Family and Patient Support section of the Rare Disease Foundation website for further information on counselling services.

http://www.rarediseasefoundation.org/family_and_patient_support

THE PATH TO EMPOWERMENT

Tips and advice to help you advocate for your child

- ☑ Focus on the current symptoms and gather as much knowledge about them as possible.
- ☑ Forgive yourself for not knowing everything. Be patient with yourself, it's hard. It's hard for everybody
- ☑ Don't believe in the limits that other people set for your child. Let your child show you their limits!
- ☑ Be persistent with your child's educators and healthcare providers
- ☑ Recognize that others are afraid too - you are not alone!
- ☑ Do the best you can: no one is perfect
- ☑ Keep your eye on the ball but don't miss the process of them growing up. You don't want to miss out on the fun and special moments because you were too stressed out
- ☑ **Positive communication** is key - stand up for yourself and be respectful to get what you need, even when people or systems seem to be completely blocking you

- ☑ Your whole job is to stand up for your kids - do not feel shy to e-mail or contact your child's healthcare providers. You have to do what's best for your kids and not worry if you're bothering or annoying other people
- ☑ You stop caring what other's think - that your child gets what they need is the most important thing
- ☑ It's pretty tough, but boy, you just get a very thick skin and you soldier on

Tips and advice for getting resources and funding

- ☑ Life with a child without a diagnosis is expensive. Sign up for each government program you can get; anything that can give you access to programs: find it, and sign up for it!
- ☑ Be persistent! It may take several rounds of applications before finally being accepted into a program
- ☑ Don't just list "development delay" - be specific. Lay out all of your child's symptoms, describe them in detail
- ☑ When you don't have a diagnosis to list, sometimes explaining the difficulties in your child's day-to-day life can be helpful for applications
- ☑ Enlist the help of doctors, nurses, genetic counsellors, and other members of your team to write letters of support for your applications
- ☑ When you're talking to people about resources, talk about difficulties openly

“By the time she was referred for any services, of course one of the questions I got was: what took so long for this child to get here?”

MAKING SENSE OF IT ALL

OK, now what? Insight from other parents

Q. Whats does the future hold?

- A. You never know how your fears are going to pan out, and you just can't handle it if you're spending all of your time thinking about the worst...you of course have to allow the grief, and you can't force the acceptance. But slowly, in time, you have to move on.
- A. At the beginning, we were only focusing on the end result and worrying about the worst.
- A. I wish I'd had a better grasp that things would progress and that we'd carry on and continue with our new life.
- A. I no longer expect the worst case scenarios (though I am well prepared for them!) Do NOT believe in limits!
- A. I deal with how it is now; I deal with everything day by day. You can't focus too hard on what might happen, because you just don't know! Anything can happen, right?



OK, now what? Insight from other parents

Q. How do I accept the uncertainty of not having a diagnosis?

A. Part of learning to live with the uncertainty, is learning to live with not always having the ‘right’ answer.

A. You have to be patient, you have to accept it. You have to realize it is there and it’s not going away. You have to work through it as it comes, and figure out the best ways that you can provide for your child... You just have to deal with it: it is what it is.

A. There are challenges with not having a diagnosis, but it doesn’t determine everything - you don’t have to have a name to be happy. It doesn’t ruin your everyday life; you could still take them to school and have fun with them. Even if you had a diagnosis, it wouldn’t change who they are.

A. There’s a lot of uncertainty, but you don’t dwell on it. Life’s about other stuff.

A. Life will never be the same - and that’s OK!

RESOURCES

The Rare Disease Foundation

Patient advocacy organization based in Canada.

Cross-disease support network: you do not need a diagnosis to be part of this community. If there is no support network in your area, set one up! Email families@rarediseasefoundation.org for more information.

Peer support; online community; links to resources.

<http://www.rarediseasefoundation.org>

The Rare Disease Foundation Wiki Resource Page Resource tool with links and descriptions of a wide range of health, educational and social services.

<http://www.rarediseasefoundation.org/wiki>

Whether you have no diagnosis, a new diagnosis or any diagnosis, join us in the global celebrations for Rare Disease Day on the last day of February every year!



RAREDISEASEDAY.ORG

Other organizations that support patients and families living without a diagnosis

Contact a Family:

Patient advocacy organization based in UK.

Their resource – Parent Guide, Living Without a Diagnosis, was the inspiration for our booklet!

<http://www.cafamily.org.uk/>

Patient Kind:

Patient advocacy organization based in Canada.

<https://patientkind.org/>

Global Genes:

Patient advocacy organization based in the United States.

<https://globalgenes.org>

Genetic Alliance:

Patient advocacy organization based in the United States.

<http://www.geneticalliance.org/>

SWAN USA or UK – Syndromes Without a Name:

Patient advocacy organizations based in the United States and United Kingdom.

<http://swanusa.org/>

NIH National Human Genome Research Institute:

Finding Reliable Information Online/Evaluating Health Information:

<http://www.genome.gov/11008303>

Financial issues

The Special Needs Planning Group:

Website with tax tips, information on setting up wills and trusts, and background on the Registered Disability Savings Plan (RDSP)

<http://specialneedsplanning.ca/index>

Disability Tax Credit:

<http://www.cra-arc.gc.ca/E/pbg/tf/t2201/>

How this booklet was made

We work in Medical Genetics in British Columbia, Canada, and help care for many people who do not yet have a diagnosis. We were inspired by the “Contact a Family” UK group (www.cafamily.org.uk/) that created a ‘Living without a diagnosis’ information resource. We first obtained funding from the Rare Disease Foundation Microgrant Program, then did our own literature research, circulated questionnaires to parents, performed structured interviews with parents, subjected the transcripts to thematic extraction, carried out focus groups with drafts of this booklet and had many health care providers and parents review our drafts. This is the result and we are always looking for ways to improve it!

Please contact us (Kelsey Lynch, Jennifer Thompson, and Millan Patel) with suggestions for improvement:

volunteer@rarediseasefoundation.org



**RARE DISEASE
FOUNDATION**

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<http://www.anothercreative.ca>

