> Not knowing what's wrong, why it happened, what's it's called - it's just terrible, I feel like we are in limbo
>
> *Mother*

Diagnosis Unknown

> They say knowledge is power and because I don't know why or what my son's condition is called, I feel so powerless to help him – what, if anything, could I be doing?
>
> *Father*

Diagnosis Unknown...

Information for families seeking a named diagnosis for their long term sick or disabled child. Finding a diagnosis for many conditions can be a long and frustrating process and occasionally it may not happen.

This booklet is designed, and is provided, for those interested in pursuing a diagnosis, where this has been a challenge. Based predominantly on families real experiences, gathered by our Nurses over many years of supporting families in these situation, and reflecting the possibilities and opportunities.

> It is believed that thousands of parents and their children go through life never knowing what named condition affects their child
>
> *Newlife Foundation*

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Why can't the Doctors tell us why our child is like this?

Why can't they tell me?

Be assured that professionals do genuinely want to find a named diagnosis for your child. However some conditions are harder to diagnose than others. It is also sometimes the case that having a named diagnosis would not initially alter the treatment, therapy or outcome for a child and so sometimes the name of the condition is not as strenuously pursued as perhaps a parent may desire. This may be because doing what is of immediate need is more important to prevent deterioration or relieve symptoms, naturally overrides the pursuit of a named condition. But you should be assured that most Doctors or specialists will want to pursue this, as having a named diagnosis can help long term and short term with treatment and management.

Regrettably it is the case, that in some rare instances, we will never have a specific diagnosis. This is because each individual person/child is a truly unique arrangement of genes and in some cases may have a condition that is not fully recognised yet. As research progresses we will continue to find new diseases and new methods of diagnosing them.

Understand that a Doctor or relevant specialist can only use the tools he has available at that time to carry out tests to try to identify a condition. Because of the number of conditions and the variety of their symptoms and the variance in which the severity can be experienced in conditions which can be inborn or develop as a result of acquired conditions, sometimes the professional may never have seen a case of a particular condition, syndrome, disease or disorder. So he or she may need to make referrals to other specialists, such as a Geneticist or a specialist Paediatrician, to help make a diagnosis.

How can a Doctor help to find out a name for my child's condition?

What can be done?

There are two basic ways that conditions are diagnosed and these can be used alone and in conjunction with one another:

- **Clinical tests – physical tests using body samples such as blood, saliva, biopsies, etc. or through images from X rays to MRI's and scans etc.**

  Such tests can take time to get the results from. Sometimes the samples need to be sent away for specialist laboratories to work with. But the doctor will usually be able to give you some idea of the time scale. Sometime samples need to be repeated if tests are inconclusive. Be patient and work with the professionals to support the tests being carried out.

- **Clinical feature recognition - assessment of symptoms gained from physical examination or assessments.**

  This may include measurements or assessments of body features. Some families feel these are less conclusive than science based testing but it is the case that some conditions are predominantly established through this method. The quality of this is in the skills of the person undertaking the assessment being experienced and thorough and sometimes this will mean a referral to a specialist centre to obtain a quality assessment. Even if this is outside your local care most families find that it is worth making the effort to attend such a specialist centre.

Remember, that while medical research has made major advances, much more investment is needed to increase the number of conditions which can be tested for.

It is important that your child attends their regular appointments even if they do not have a formal diagnosis. Keeping track of changes might offer clues to a diagnosis.

My child was tested when he was young but they didn't know what was wrong then. Could they tell me now there are new tests?

Is it worth trying again?

Many tests have developed greatly over the past few years, especially genetic and chromosome testing.

Therefore, those children who have had tests, which were carried out over 10 to 15 years ago, could be considered for re-testing again using more recent techniques. There are no guarantees but there is little to lose in trying.

First step may be speaking to your GP or asking for a referral to a Paediatrician or Geneticist.

I always thought my child was this way because of something I did – I have always felt guilty about this – could having a diagnosis help?

Why did it happen?

Almost every parent of a child with an inborn condition or acquired condition asks, "why?"

It's perfectly natural to be concerned about why a condition occurred and it's natural to want to find out some answers. Pursuing a diagnosis may or may not achieve a specifically named diagnosis, but a specialist may be able to clear up this concern for you about cause.

Guilt can be a very damaging emotion, so it is not unreasonable to explain that this is your concern. Most families, in time, come to realise, that while they may never know the cause, they did not intentionally do anything that caused this condition so responsibility for the condition does not lie with them.

There is much mythology that haunts parents when considering this issue and these are not helpful. Remember that myths are made up to try to explain things that people don't understand. It doesn't mean they are true, but many families find such sayings and old wives tales very hurtful and unhelpful. Of course rationally we know this is not the case but pursuing a diagnosis can help blot out such myths and difficult feelings of guilt.

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The queries shown below are all questions received from real parents who have real concerns about what could have caused their child's condition:

"I was on a diet before I conceived and I didn't know I was pregnant for a while so I wasn't eating well in the first few weeks"

"My husband worked in an industry where there have been rumours that health is affected - could this have caused this condition?"

"I unknowingly used an insecticide while pregnant - could this have caused the condition?"

"My grandparents were first cousins"

"My child was conceived out of wedlock"

"I had a cold during pregnancy"

"My husband smoked while I was pregnant"

"I had two sherries at Christmas while pregnant"

From practical experiences which concern parents, about why the condition occurred, to cultural and religious perceptions, which can have equally powerful influences, all can cause unnecessary worrying and guilt by parents. Most parents find that by openly asking about these issues and raising their concerns, relevant professionals can clear up these matters and help overcome negative feelings about these worries and concerns. In pursuing a diagnosis, answers to these sorts of questions can also be responded to. Don't worry - medical professional's hear questions like this all the time - they won't be shocked or surprised and will be sensitive to your queries if you are clear about how much the issue concerns you.

> *I wake up thinking what will we do if and when she is a teenager, what will we need to plan for and hundreds of other worries. Having a diagnosis may help us plan and worry less.*

Does everyone wonder and worry?

Yes, everyone in this situation worries, because uncertainty about the future is very common when there is no diagnosis for a child.

It's hard to 'move on' and plan your family, when you don't have any idea of how a condition may progress or improve.

Getting a diagnosis may not fill all these information gaps but it may help. With or without a diagnosis, all parents/carers of children with disabilities face the same problems of service provision etc, but also the joys shared with their "special" child.

"When people ask me what he or she has, I never know what to say – no diagnosis or named condition. Can you suggest something in the meantime, long term, or if I decide not to pursue a diagnosis?

How do I explain?

It is so difficult not to have a specific named condition so many families find it easiest to explain what the issue is in shorthand terms.

Parents adopt various terms to do this, here are some examples where parents will say he or she:

"has a medical condition."

"has a disabling condition or is disabled"

"has a problem with xxxx"

"undergoing a test for xxxx"

"having difficulty with xxxx"

Or come up with some other term, which you feel can quickly sum up that your child needs some special provision or response from people or services. Example:

"he has an inborn genetic condition that affects his muscles and coordination".

"My child behaves this way because of his condition".

Most parents find this sort of short cut helpful and it avoids having to go into personal details. If further questions are asked most families find a reply like "the specific condition is currently un-diagnosed" usually works.

"Will my child be negatively 'labelled' if I find a specific condition name for the disorder or could I learn something I didn't expect?"

Could a diagnosis be a negative thing to have?

First let's address 'labelling'. This concern was a reality maybe 30 years ago, when the understanding of inborn and other conditions, illnesses and disabilities was poor. But no child these days would be automatically assessed for services or provision, just on the basis of only having a named diagnosis. This is because standards, understanding and appreciation of the whole and specialist needs and abilities of the individual person have greatly improved since then.

These days all care plans, education and treatment is solidly based on detailed individual assessments. Even children with the same named condition can have dramatically different care or support, as the individual traits or features of the condition can be unique to that child. So, most families feel that having a named diagnosis is a positive activity, which vastly outweighs the possibility of being concerned about labelling or being stigmatised.

As to whether you could learn from a diagnosis something that you would prefer not to know, well this is about what you tell your doctors or professional you want to know. If your concerns are about for example, future mobility, then this is what they will predominantly communicate to you. However, if you do not wish to know about more challenging information such as longevity, etc. you can be clear with the people you deal with that this is not your concern and interest.

If you change your mind later then they will be able to advise you again. In most cases experienced professionals are very sensitive to deliver information that is relevant the questions families have, they don't just 'dump;' un wanted information on you, so it will in the main be in your control by speaking and being honest with those you are dealing with.

"Will knowing a specific disorder name change how I see, appreciate or treat my child?"

Will a diagnosis change how I see my child?

Most parents say that knowing what a named condition is helps them. As one mother sums up "I needed to know, what? and why? and, when I found out, I was able to use the information where it was useful, but most of the time I just think of Susie as Susie, not Susie with this syndrome.

This is the most common experience of families. They feel the benefit from a diagnosis, but once they have learnt about it and understood and know when and where to use the proper name of the condition, they just put it aside and treat the child as they did before, but with greater insight into why some things are the way they are, and most families report that this helps them to cope better.

Often by the time parents finally get a diagnosis, they find that the label no longer matters. They themselves have already done all the hard work in finding out about their own child's "individual" needs. They often become the experts.

How can I do this?

If your child is under 16 and you have a Paediatrician, you could ask them to review the diagnosis. This could happen at your next routine appointment. He or she can pursue these themselves, carrying out tests and assessments, or they could refer to other specialists including a Geneticist.

If under 16, but you do not regularly see a Paediatrician or over 16 and not seeing a Paediatrician, you could ask your GP for a referral to a Paediatrician if you want the diagnosis reviewed.

You should explain to the GP or the Paediatrician why you want the diagnosis reviewed. Explain that you want to understand whether or not the condition could be genetic and as new tests are now available which were not possible when the child was younger, you feel it is time to pursue this.

Explain that you are concerned about why the condition has occurred and be honest and explain that this troubles you and you feel you need some answers.

You could also explain that you have concerns for the child's future reproduction options and for the reproductive options of brothers and sisters, etc. in case it is a genetic condition.

You can explain that you feel that knowing what the condition is would help you manage and feel more able to cope, not only with the day to day practicalities but in planning care for the future. Given such well explained reasons, most Doctors will feel able to make a referral in pursuit of a diagnosis.

What can I do to help the Doctors or Specialist find a diagnosis?

Can I help provide information?

Before being referred to a specialist to pursue a diagnosis there are things that you could do to help. Ask the person referring you if these may help:

Family History

Look and note brief details of your family history and briefly record any instances of repeated miscarriages, stillbirths, infant deaths, long-term conditions. Look at parents, maternal and paternal grandparents, brothers and sisters of the child, other children of you or your partner, the brothers and sisters of you and your partner.

Write down any experience/conditions and explain these in the context of the relationship to your child (not the relationship to you/your partner) e.g. if your (as mother) had a brother who had a similar condition it would be the child's maternal uncle. If it was on the father's side you would describe it as a paternal relative.

Remember, some conditions you may think are important are not, because they are called 'late onset conditions' occurring in aged relatives so these do not need to be noted e.g. a grandfather may have died of a heart condition which only became a problem when he was aged. However, if the person suffered from a heart condition from being a child, this is worth noting.

It is probably better to include all information so the Doctor can discount it, don't include hearsay from a distant relative.

Always be honest about genetic relationships, e.g. if the child is adopted or if the family history of one parent is unclear, this can occur because of adoption, etc. This information can have a significant effect on the ability to diagnose or not.

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What you know and see

Before the appointment you could also write down what you see or believe are the problems or symptoms in the form of a list, copy it and give it to the Doctor(s) concerned. This should be written as an aid to the appointment and to help you remember everything, but do write down significant issues. He or she does not need to know about every cold or measles.

Try to break these down into sections such as:

- Organs and digestion - issues with digestion, urination, bowels, heart, liver, kidneys, feeding, genital problems, etc.
- Skin, hair, nails and teeth - issue with skin tissue, healing, bruising, hair growth and loss, teeth development and loss, etc.
- Joints and muscles - mobility, stiffness, pains, stability, injury issues, co-ordination, restrictions, stamina on exercise, etc.
- Senses - hearing, sight, touch, taste, smell - lack of, excessive, poor, restrictions, etc.
- Pain - if pain follows activity or incidents, time of day, etc.
- Sporadic incidents - if the child appears to lose consciousness or has episodes of seizures or periods of absence, or other occasional issues, etc.
- Mental and emotional development, behavior and communication - perhaps the hardest of areas to describe, as most of this is relevant to the age of the child at the time. Try to centre on learning ability, milestones achieved or not, speech and ability to understand, ability to make decisions, ability to avoid injury or be safe. Excessive dependence, not acting as one would expect for age. Responses to others, outbursts. Whilst telling the Doctor how these behaviors make you feel may help you, it may not be the best way to help him/her see the practical problems and symptoms, so focus on reporting what you see rather then what it makes you feel.
- Other symptoms - these may include things like unusual sweats, smells, perspiration, soreness, tremors, habits such as repeated blinking or picking at things, obsessions, repeated infections, serious injuries and infections or surgery, etc.

I don't want to get my hopes up but I hate living without knowing but what if I am disappointed?

What can I hope for?

The minimum you can hope for is that professionals will listen to your request and understand that it is important to you. Be prepared to listen to their opinion too. They may be able to help you deal with these issues without pursuing a clinical diagnosis for a named condition.

This may be enough for some families. but if you still feel pursuing a named diagnosis of a condition is important to explain this to them and ask them to support you, Make them research if there is a specialist centre or specialist service that could help. You can ask for a second opinion but this is not guaranteed to provide further information or a diagnosis.

It could be the case that your doctor will be able to assure you that having a named condition will not make any difference to the treatment or management of your child's health or welfare. In such cases many families feel it is reasonable to accept this and feel better that they have raised the questions, explained and explored their feelings about this and then adopting a short cut to describe their children's situation - see further information below.

The old saying is that if we never try we never fail, but we also never succeed. So decide how important this is to you. If everything, after all has reasonably been done, no named condition is possible, at the very least you will be able to say you really tried and you were supported to explore this and know you have done everything you can. Don't under estimate the great comfort this can be on a daily basis, because doing what is the best for your child is after all what is most important.

Can anyone help me decide and act?

Yes, Newlife Nurses can help you, Our 'Diagnosis Unknown' service is based on a freephone Nurse helpline where your can speak in complete confidence, discuss things that you wouldn't say to others but need to know, and that it is okay to ask or want to know more information about.

Newlife nurses are experienced people who have experience in children's health and disability. So they know the issues, they care and will not judge and can give you information so you can make an informed decision.

We understand the frustration of not having a diagnosis and in fact Newlife's co-founder had to wait until her son was 9 years old to get a diagnosis so we are very aware of these issues.

About Newlife Foundation

The UK's leading disabled children's charity

Disabled children have been at the heart of the charity since 1991. Since that time we have become, though our work, the specialists for special children. Their needs and the needs of their families and carers, their health and their voices are all central to the work of the charity.

Originally the charity focused on the biggest cause of disability in children, inborn conditions. As times have changed and more babies and children survive conditions, accidents and infections and cancers . we see more children thankfully live through these experiences. However as a result many go on to face both short term and enduring disability. Others will live with their life expectance limited, while tragically others children become terminally ill. Newlife exists to respond to all these children with care, professionalism and determination and to provide informed support to their families and carers.

Newlife is a national charity and has 3 key areas of activity:

Nurse Care & Equipment Services

Nurses and specialist equipment making a real difference to the lives of children and families.

Medical Research Action

Pioneering work to improve child health, treatment and the knowledge to prevent disability.the best researchers provide the best chance of improving child health.

Campaigning & Awareness

To achieve improved services and knowledge and to be the voice of affected children and families.

All services are free to families;
we do not 'means test'.
The needs of the child are paramount.

Social Media Action

If you're a fan of social media - either personally or corporately - tell your followers all about Newlife. Or show your support for us through advertising; River Island, for instance, outlines the charity's aims on the back of all its till receipts and includes our Newlife logo on its carrier bags.

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