



helping
to
cope -
helping
to
hope



ALD LIFE
45 PECKHAM HIGH STREET,
LONDON SE15 5EB

Tel: 020 7701 4388
www.aldlife.org
info@aldlife.org

Issue Date: January 2015
Review Date: January 2016

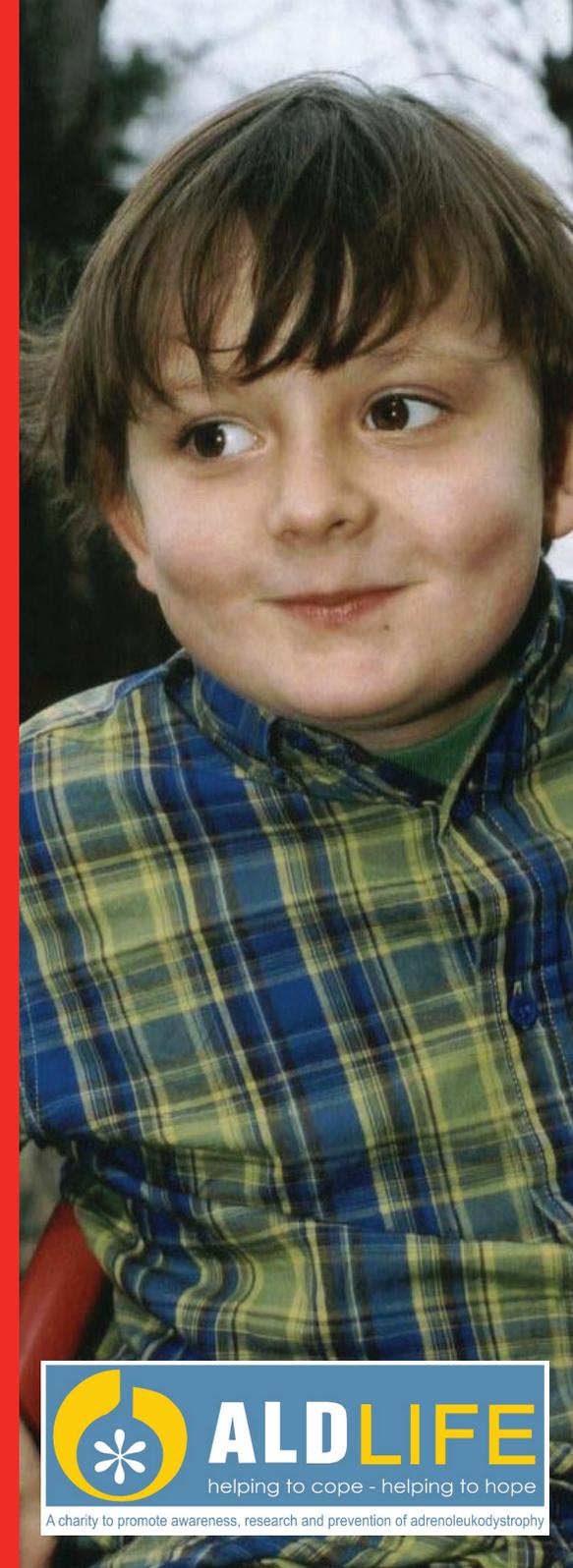
registered charity number 1106008



PRACTICAL INFORMATION
FOR MALES WITH
SYMPTOMATIC ALD



A charity to promote awareness, research and prevention of adrenoleukodystrophy



PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Introduction

YOUR child or relative has been diagnosed with symptomatic cerebral adrenoleukodystrophy (ALD). It is an extremely rare genetic illness that affects boys – mainly aged four to ten – but can also affect male teenagers and adult men.

Living with a chronic disorder like ALD is very difficult but you are NOT alone.

ALD Life is Britain's leading charity dealing with this condition, run by people who have been in the situation you are now facing and who can help you.

We were founded to help and support people in your position. Our members have been through the same ordeal and have experienced the problems you now face.

We have a worldwide contact list of people who are willing to chat about all aspects of living with the disorder – from being a carrier to having lost a child. Most contact is done through email, one of the easiest ways to talk to people as it is less emotional. We also have forums on our website.

Please contact ALD Life on **020 7701 4388** or email **info@aldlife.org**. Our website is **www.aldlife.org**.

You are going to become the best advocate for your child or relative, because you will know him and his condition inside out.

You are going to have to learn how to become a nurse, a physio and an expert on ALD.

A whole host of professionals will tell you what they think is best. You will have to learn to scrutinise what they say and decide what is right for your family.

You will be the ones who make the decisions. You will have to very quickly learn how to weave your way through the maze of health and social services to get the help you need.



PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Our members have helped compile this leaflet, which is packed with information other parents in your position wish they had been told.

In researching this booklet, we have spoken to many parents whose sons have symptomatic cerebral ALD. Much of the information is therefore based on experience. The medical information, presented in shaded boxes, has been carefully compiled from reputable sources (listed on p.27) and has been reviewed by ALD specialist doctors.

What is ALD?

Adrenoleukodystrophy (ALD) is thought to be caused by a genetic fault that means those affected are unable to process Very Long Chain Fatty Acids (VLCFAs). It is thought that these VLCFAs accumulate and destroy the myelin sheath that covers the nerves in the body and brain. The myelin acts like the coating around an electric cable, and allows messages to be transmitted along nerve cells.

The gene at fault in ALD is called ABCD1. The faulty gene can cause several related but different conditions. This leaflet is about symptomatic cerebral ALD, which is the most devastating form of the disease. We have another leaflet on asymptomatic ALD (for males affected by the gene who do not yet have symptoms). The other condition caused by the faulty ABCD1 gene

is called adrenomyeloneuropathy (AMN), which affects nerves in the spinal cord and body (see our AMN booklets for men and women).

In cerebral ALD the damage to myelin happens in the brain. When the myelin is damaged the nerves in the brain cannot work properly, and the person's physical and mental abilities begin to deteriorate. Functions such as reasoning, speech and mobility are lost. Eventually, they become completely dependent.

In most people with ALD the adrenal glands are also affected. The adrenal glands make steroid hormones. Poor functioning of these glands is known as adrenal insufficiency. Symptoms include weakness, tiredness, vomiting and muscle pains. It is not possible to predict how the ALD gene will affect any one person: its effects can vary. Once diagnosis has been confirmed, the affected person needs to be closely monitored.

How is ALD diagnosed?

Initial diagnosis is usually based on an MRI scan and a blood test for Very Long Chain Fatty acids (VLCFAs). High levels of VLCFAs, along with white matter deterioration in the brain, are good indicators of ALD being present. Diagnosis is confirmed by a genetic test. The genetic test takes some time – as much as six weeks to come back.

Once the genetic tests have confirmed the diagnosis, immediate family members should be offered urgent genetic testing.

There is more information on p.8 about genetics and ALD, including why family testing is so important.

Can ALD be cured?

Sadly, once symptoms of cerebral ALD have appeared, there is currently no cure. However, it may be possible to prevent the disorder in people who carry the ALD gene but have so far developed none or minor symptoms.

Medical guidelines say that on average, boys with symptomatic cerebral ALD live for 2-4 years. But many boys have gone on to live for longer periods. There are boys who have symptomatic ALD that are living in their teens and early 20s with quality of life.

My son has just been diagnosed with symptomatic cerebral ALD – what should I do?

It is the most devastating news anyone can imagine. We must stress there is no right or wrong way of dealing with this news. However, this is precisely why ALD Life was set up – to offer support and advice to those facing the isolation of a symptomatic ALD diagnosis.

“ IT WAS A RELIEF TO FIND A RELIABLE & TAILORED INFO CENTRE LIKE ALD LIFE ”

“ DON'T ALLOW YOURSELF TO BE INUNDATED WITH PROFESSIONALS JUST AFTER THE DIAGNOSIS - ONLY SEE PEOPLE WHO CAN HELP YOUR CHILD AT THAT PARTICULAR TIME. ”

“ HAVE FAITH IN YOURSELF - YOU KNOW HIM BETTER THAN ANYONE ELSE. ”

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

The rest of this leaflet offers practical advice from parents who have been through this experience, and gives more information about ALD and how to access the support you need from health and social services.

What do I tell my child or relative, family and friends?

What you tell your child or relative, your family and friends is your personal choice. You know them best and know what they can and can't take.

Some people say honesty is the best policy but others will delay telling the patient what exactly is wrong with him for some time. Sometimes it may help (with your permission, of course) for a member of the medical team to break the news.

With children, parents may find it best to start simply by giving the illness a name and then explain later that this condition has to get worse before it gets better.

Our experience is that most children seem to take what you are telling them quite matter of factly. With all things like this, kids are fantastic and adults struggle with it! Children can be incredibly resilient.

One of the slight blessings of this condition is that the patient experiences dementia-like behaviour so it is likely he won't fully comprehend what is happening to him. So, you do not need to let them

know the full extent of what is going to happen. And because of the illness, he will not take in everything you are telling him.

There is a lot happening at this time and dealing with the reactions of other people can sometimes be quite traumatic, so you may want to tell a few friends and let them spread the word to other people.

Although things won't necessarily get better, many people find that with time, the situation does become easier to live with.



Finding more help

Together for Short Lives is a charity set up to help children who may not reach adulthood.

Their website is packed with useful information for both parents and professionals.
<http://www.togetherforshortlives.org.uk/>
call 0808 8088 100

“
I TOLD MY SON HE HAS
AN ILLNESS THAT NEEDED
TO GET WORSE BEFORE
IT GOT BETTER.”

What about schooling?

You will need to talk to the school about continuing education.

How schools cope with ALD sufferers varies greatly. Some will be very supportive, while others may find the situation too difficult to cope with.

When a child with symptomatic ALD starts to deteriorate, it can be quite traumatic for their school friends, who have probably never experienced anything like this before. In fact, it will probably be a whole new experience for the school.

We hear wonderful stories of some mainstream schools that will keep a child there for the whole time, but usually they will need to move on to a special needs school.

Again, it is an individual decision. Some people want to pull their child out of school as quickly as possible and keep them at home all the time, while other parents want their child to stay at school and be with their friends for as long as they possibly can.

Why is genetic testing of the family important?

Once one member of a family has been diagnosed, it is very important to get the rest of your family checked genetically, especially if there are brothers, who could also have the disease. If the gene is discovered early, before symptoms appear, there is a chance of preventing ALD from developing. Sisters could be ALD carriers.

When a boy is diagnosed with ALD it will most likely be inherited from his mother.

You should be offered genetic counselling as soon as the diagnosis has been confirmed. Genetic tests are advised for all siblings and parents. The genetic counsellor will talk you through the implications of the tests.

If siblings are found to have ALD they should be considered for a preventative programme, which is explained in another leaflet - Males with Asymptomatic ALD.

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Explanation of the genetic implications

ALD is an X-linked disorder, which means that the genetic abnormality involves the X-chromosome.

Women have two X chromosomes. In women who carry the ALD gene mutation on one X-chromosome, the full-blown disease does not appear because there is a normal copy of the gene (Figure 1) on the other X-chromosome.

Men have one X-chromosome and one Y-chromosome (Figure 2). In men who have ALD on an X-chromosome there is no other X-chromosome for protection; therefore the male can develop symptomatic ALD.

For reasons we don't understand, not all males who have the gene are affected in the same way. Some develop cerebral ALD as boys, others have no symptoms for many years and then develop AMN as adults. Some males develop Addison's disease (adrenal failure) rather than ALD or AMN. Most women with the gene will also develop some, usually mild, AMN symptoms later in life.



Figure 1:

If a woman is a carrier for ALD she has the following possible outcomes with each newborn: With a daughter, there is a 50% chance (1 in 2) that the daughter is a carrier of ALD and a 50% chance the child is unaffected. Where the child is a boy there is also a 50% (1 in 2) chance the son will have ALD and a 50% chance he will be unaffected.

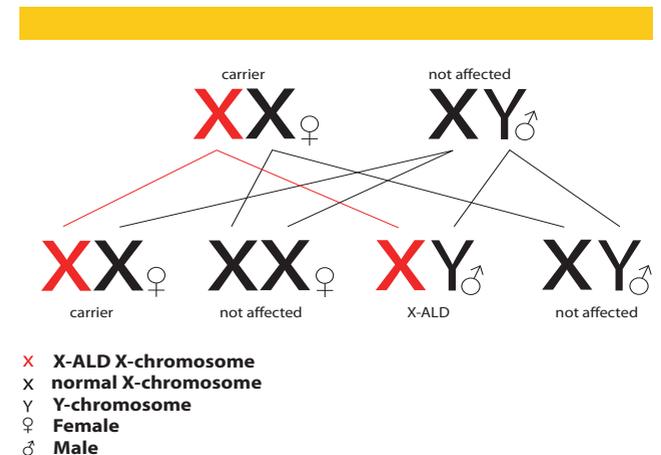
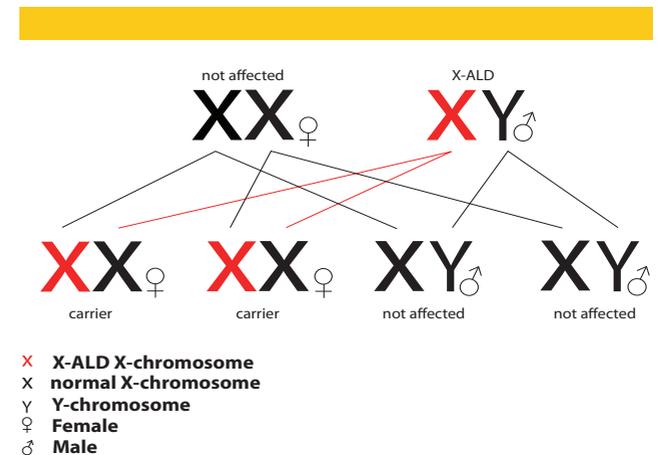


Figure 2:

If an affected man has children, then all of his sons will be entirely normal (because sons get the father's Y-chromosome). But all of his daughters will be carriers (because he passes his only X-chromosome to his daughter).



PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

What medical support do we need?

The diagnosis of symptomatic cerebral ALD will probably be made by a doctor specialising in metabolism, the body's chemistry. ALD is classed as a 'metabolic disorder'. You will probably not see the metabolic specialist again after diagnosis. This is because they are only usually involved in the prevention of ALD before symptoms progress. Unfortunately they will not be able to treat your child.

The patient may also be diagnosed by a neurologist, who will keep an eye on his condition. The neurologist is the person you will have most contact with, as they will look after the various symptoms associated with ALD.

You will also be visited at home by a district nurse, who will be your link to Community Health Services in your area. They will handle everything to do with medical needs and equipment in the home.

It is important that you have a good relationship with a physiotherapist and possibly the orthotics department at your local hospital. Orthotics are the people who will provide you with equipment like splints and braces.

How quickly will he deteriorate?

The deterioration rate is different for each patient. Some will continue to walk and talk several years before succumbing to the effects of the disease, and will then suddenly deteriorate very quickly into a dependent state. Others will deteriorate into the dependent state within a few months of diagnosis. There is no way to predict how quickly or slowly the process will happen.

For a very small minority the disease can stop spontaneously in its tracks. It is still unclear why this happens.

It is a cliché but you have to take one day at a time and see what happens.

What symptoms will he get and how will they be dealt with?

Adrenal failure and steroids

Once the patient has been diagnosed with symptomatic ALD, make sure he is immediately checked for adrenal insufficiency. In ALD the adrenal gland, which produces an essential hormone called cortisol, can stop working. The consultant who made the ALD diagnosis should refer the patient to an endocrinologist, who will be responsible for making sure adrenal tests are carried out.

Adrenal insufficiency can be treated easily with steroid tablets (hydrocortisone and fludrocortisone) to replace the hormones

the adrenal gland usually produces. If untreated, adrenal insufficiency can lead to a life-threatening condition called 'adrenal crisis'.

A common sign of adrenal insufficiency is being bronzed, i.e. becoming progressively tanned even in winter, sometimes with brown creases on the palms of the hands (rather than red ones). Other signs include becoming lethargic and having difficulty recovering from infectious illnesses.

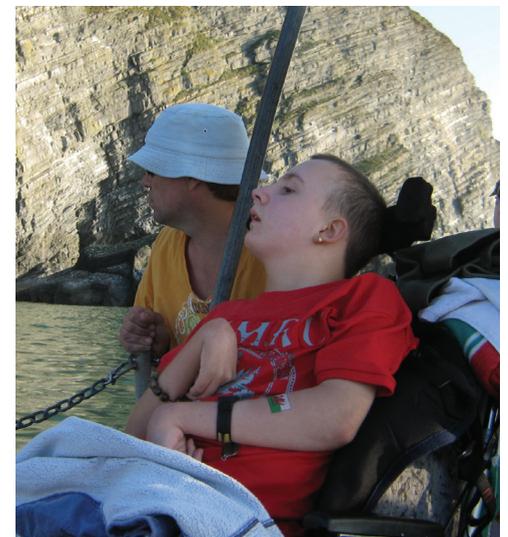
Another possible sign is 'cyclical vomiting', where the patient catches a bug, starts to vomit and quickly looks very ill and has to be admitted to hospital. Once there they are quickly 'cured' by hydration and often discharged the following day.

On very busy or stressful days patients with symptomatic ALD may go through their steroids faster than normal, causing tiredness, vomiting, headache or fever. Some patients get hangover effects from busy days where they have to stay off work or school or go home early the following day simply because their system has become overstretched. Also, you will be given advice to increase the dose of hydrocortisone immediately the patient becomes unwell or is prescribed antibiotics.

You will also be trained to give an emergency cortisol injection in case the patient is not retaining his hydrocortisone due to vomiting or diarrhoea.

The endocrinologist should also give you a letter stating that if the patient has to go to Accident and Emergency they are to be seen immediately because they suffer from adrenal insufficiency. The endocrinologist will also inform the ambulance service that the patient has adrenal insufficiency so they carry the correct medication for him if you need to call 999.

All with adrenal insufficiency should have a medical identity bracelet or necklace stating their medical condition that they wear all the time. That way if something happens and the patient is alone, medical authorities will be alerted to his condition and be able to treat him properly. There are many places now that supply these - just search online for medical ID jewellery for a list of suppliers.



PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Muscle spasms, fits

Males with symptomatic ALD face many of the same problems as any patient with chronic disabilities. Muscle spasm is a typical symptom of ALD and can be very painful, so the patient will need regular physiotherapy and sometimes body bracing and splints for the feet.

It is very important to keep the muscles moving to help with muscle spasms, so you will need to learn to give physiotherapy. There are also drugs that can be taken for muscle spasm – your neurologist will help with medicines to treat these symptoms. In extreme cases botox injections can help.

Having fits is another problem males with symptomatic ALD may face. Some have a lot of fits while others don't have many. Anti-convulsants are a medical option for this problem.

Chest infections, pneumonia and vaccinations

Chest infections are common and patients with symptomatic ALD are also at risk of pneumonia. However you may be able to reduce this risk by ensuring that your son is given a vaccination against pneumococcal germs, which can cause pneumonia. He should also have the annual flu jab. Some boys may also benefit from regular preventative antibiotics during the winter if they tend to get recurrent infections. Also, please try to make sure that you, the rest of your family and your helpers all have the flu jab too.

A lot of families are never told about the dangers of over-medication. You need to be aware that medications that cause drowsiness can also mask the symptoms of chest and viral infections, which can be fatal if they develop.

Continence

Eventually the patient will become incontinent; with the help of an incontinence assessor from your Community Health team, you will need to look at options to deal with this. Alternatives are using pads (nappies), sheaths (usually preferable as they are drier and free from possible soreness) or catheterization (more usual for adult males, but can carry a risk of persistent urinary infection).

The community health team will organise things like regular delivery of incontinence products to your home.

Swallowing and feeding

At some point the patient will deteriorate to the point where he will not be able to talk, and he will not be able to swallow properly. At this time you will need to start thinking about feeding tubes and alternative methods of communication.

There are several forms of tube feeding and the Community Health Service will guide you through the options. Most males with symptomatic ALD will eventually have a gastrostomy put in. This is a small

operation to insert a feeding tube directly into the stomach. The alternative is a nasogastric tube, but most people tend to prefer gastrostomy because nasal tubes have to be changed often and can easily be pulled out.

Your Community Health team will manage this type of feeding and will produce a feeding regime. They will also monitor the patient's weight and make sure he receives the correct nutrition and the right amount of water.

Some males develop chronic wind problems when they start tube feeding. This is not easily treated through medication and many try different options such as positioning, natural remedies and opening the gastrostomy tube to release excess air. A device called a Farrell valve can also be used to assist with this problem during tube feeding; many report that this is very effective.

You can still feed the patient orally while they have a feeding tube. This will be monitored by a speech and language therapist, who can also help with communication. Saliva control and the build-up of thick mucus secretions can be a huge issue in the care of someone with symptomatic ALD. There are various methods that can assist with this from medication to physiotherapy and, for more severe cases, botox and radiotherapy. A physiotherapist and/or speech and language therapist will be able to advise on the best treatment.



“ TRY NOT TO BE INTIMIDATED BY MEDICAL PROFESSIONALS. ”

“ PNEUMONIA IS PREVENTABLE, BUT THEY NEVER TOLD ME THIS UNTIL AFTER MY SON GOT IT. ”

“ WE WERE CONCERNED THAT HE WASN'T GETTING THE BEST NUTRITION THROUGH HIS GASTROSTOMY SO WE BOUGHT A JUICER AND GIVE HIM FRESH VEGETABLE AND FRUIT JUICE EVERY DAY THROUGH HIS TUBE. ”

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

What happens if his brothers are found to have ALD?

If brothers are found to have the ALD gene they should be considered for a preventative programme, which is explained in another leaflet - Males with Asymptomatic ALD.

What about mobility?

As deterioration continues, you will be contacted by an Occupational Therapist who will look at your housing situation, whether your home is suitable for adaptation or whether you need to perhaps move house or build an extension, depending on your circumstances.

It may be that you will have to have a lift installed. Stair lifts are not usually recommended because of the muscle spasms. Grants to have your house adapted are available.

Because the patient's ability to move will deteriorate, he will eventually reach the point where he will not be able to bear his own weight. There will come a time where you will need an electric-powered hoist for him to be moved into the bath, into bed or into his wheelchair.

You will also need an electric powered fully-profiling bed, which should be provided by Community Health. Or there are grants available from various charities to help you purchase one if you prefer.

Because the patient will be immobile, you need to be able to help him change position in bed to help his breathing and to avoid pressure sores. Community Health will also be able to provide special mattresses.

How do I get help with care?

Looking after a disabled patient is very exhausting and you simply CANNOT do it all on your own. It is important for you to have a life yourself. You would not normally be with your loved one 24 hours a day.

It is also important for their social and psychological wellbeing that they are introduced to other people.

The rest of your family will also still need to be looked after. If you have other children they will feel neglected if you don't devote some of your time to them, and your home will still need to run.

Some families don't need outside help if they have a network of family or friends to call on. If you are not in that position, you need to turn to social services, and make sure they provide a care package where people come in to your home to help you look after your loved one with ALD.

Your helper can have whatever role you require. Perhaps getting the patient up and looking after him while you get your other children to school. Or perhaps providing cover so you can get time to go out during the day, or helping to feed him in

the evenings. Maybe the patient regularly wakes in the night and you need someone to be with him, so you can get a good night's rest.

Once you have been allocated helpers you will need to learn how to cope with having strangers in your house.

“
YOU NEED TO LEARN TO BE FORCEFUL IN ASKING SOCIAL SERVICES FOR THE HELP YOU NEED.
”



You need to learn how to effectively ask social services for what you need and how to reject unsuitable people. You are the one in charge of looking after your loved one. You have to have a team you are comfortable with and who will work to the system and methods that are best for your family.

One of the key things to make social services realise is that if they are going to give you a care package, it has to be made up of people who come to help you regularly and have a vocational interest in looking after the patient. Both you and your loved one need to feel comfortable with them. Some helpers are fantastic, but some are not.

The process of getting the right people in can take some time, and there may be a period when it will be difficult. Be persistent in dealing with the situation and you WILL get the right care eventually.

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Respite care

There are hospitals and hospices where your loved one can go to so that you, your family and the patient can get respite for a week.

You can either contact your local hospice direct and refer yourself, or you can ask your social worker to refer you.

Respite care does not suit every family. It has to be a personal choice, but it's an option you should perhaps try to give you, your family, and your loved one a break. We all fear something untried and untested, but once they have tried respite care, many wonder why they had not done it sooner.

There are other options, for example you can ask to have a clause written into your care package that allows you and your family to go on holiday each year with a carer to assist you.

Getting out and about

Having a loved one with a chronic disability like symptomatic ALD does not mean you have to stop doing things as a family. The more you do the better, because it helps the family as a whole and gives your loved one a sense of normality and vital stimulation to keep going.

There are such a lot of things that you can do and places you can go with an ALD patient in a wheelchair. More and more activities are now accessible to those with chronic disabilities, like going bowling, to the cinema or swimming, and adapted equipment can now be found in places such as your local park to enable those in wheelchairs to enjoy playground equipment. These days there are many options for accessible holidays complete with hoisting equipment and hospital beds both at home and abroad.

Males in wheelchairs don't have to miss out on adventure holidays, where pursuits like zip wiring, abseiling and canoeing are not only possible but actively encouraged. The Calvert Trust, www.calvert-trust.org.uk



for example, runs three centres, in the Lake District, Northumberland and on Exmoor, where disabled youngsters can go on individual or family holidays to experience the outdoors.



How do I give him quality of life?

We know just how hard it is when your loved one is diagnosed with symptomatic ALD, but it is so much better for him if you can stay positive.

At the beginning a good piece of advice is to try and keep life as normal as possible. Don't stop telling your loved one off, or start giving them everything they want, or they will become suspicious that something is wrong. As quickly as you can, do go for the trip of

a lifetime, before further deterioration sets in. On diagnosis you should be given details of organisations such as Dreams Come True who can help with this.

“ WE TRY TO DO EVERYTHING WE USED TO WHERE WE CAN AND KEEP THINGS ‘NORMAL’. NO SPECIAL TREATMENT AND WE INSIST THAT HE IS NOT BABIED OR TALKED DOWN TO. ”

What about my other children?

It is often hard for brothers and sisters to continue life as normal and accept what is happening. It is normal for siblings to have immense feelings of guilt about the situation, and to feel neglected by their parents.

Counselling is always available at the hospital for both children and adults. Play therapy and other forms of counselling to help children and adults deal with what is happening can be offered. The older a child gets the more difficult they tend to find it, as they become more aware of what is happening to their sibling.

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Some families find organisations such as the Young Carers Project www.youngcarers.net can help them, as they introduce siblings to others in similar situations and arrange special outings for them.

What am I entitled to?

On diagnosis of symptomatic cerebral ALD the hospital should also be the people who start the process of getting you Disability Living Allowance (DLA) or Personal Independence Payment (PIP). They will also put you in touch with social services, and you should be assigned a social worker immediately.

Although people are supposed to come to you and offer you the services you need, we find from experience that it just doesn't work that way anymore. You need to be very pro-active in asking for the help you are entitled to.

Our guide lets you know what you should be offered and how to go about getting it.

In theory, once someone has been diagnosed with symptomatic ALD, people from social services will come round to see you to talk about the benefits you are entitled to. They should also liaise with Community Health and Occupational Therapy to ensure the correct services and equipment are installed for your loved one's needs.

Grants and benefits

There are various grants available for people with disabilities. The **Family Fund** www.familyfund.org.uk is a charity that helps families with severely disabled children and gives grants for things such as washing machines, driving lessons, costs of hospital visits, computers and holidays.

There are also many other charities that will provide money for equipment and holidays (see our list on page 20). Also look out in your local library or online for **Grants for Individuals in Need**, which has proved to be invaluable for those looking for charities who have funds to help out.

The Directory lists every charity in the UK and how they can help people like you. Some charities require a referral from your social worker but many will accept a letter from you setting out your case.

Our charity ALD Life provides grants to families who have gone through a bereavement as a result of ALD or who have a child going through a bone marrow transplant. We also provide grants for equipment & therapy that is not available on the NHS.

Everybody with symptoms of ALD should be receiving Disability Living Allowance (for under 16s) or Personal Independence Payments (for those over 16) at the

appropriate rate. If you have an asymptomatic boy on steroid replacement you are entitled to the lower care rate. Symptomatic males will usually qualify for the highest rate for care and mobility.

As a carer of a family member with symptomatic ALD you should receive Carer's Allowance.

For major adaptations to your home your local council should assist if you are in council accommodation. For those in housing association properties, the landlord should help but is not legally obliged to. If they will not help you, approach the Home Improvement Agency, who also provide grants for privately owned properties.

You can apply for a wheelchair adapted vehicle from the motability scheme www.motability.co.uk

For small items of equipment you can apply to various charities for funds. Your social worker or occupational therapist should be able to provide you with a list or you can source funds yourself using Grants for Individuals in Need.

The charity Contact a Family www.cafamily.org.uk are very good for those with disabled children – they will even make home visits to go through with you the benefits you are entitled to and look at your housing needs to make sure

you are receiving the financial help you need. **Free helpline 0808 808 3555.**

Community Health services should provide you with a lifting hoist and an adapted bed and mattress when you reach the stage of needing them.

Adults with symptomatic ALD

On reaching 18 your loved one will go on to adult services and all his medical, educational and social needs will be met by adult provision. This transition can be stressful, so try to ensure everything has been assessed and put in place before his 18th birthday.

If you are already an adult who has progressed to having cerebral symptoms, your options for care and medical needs are very similar to those of boys with symptomatic ALD but will, of course, be met by adult services.

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Bereavement

You can expect your health team to be open and honest with you when they believe the end of your loved one's life is approaching.

You will want to make the most of the last weeks and days of your loved one's life and spend as much time as you can with them.

Meanwhile, your care team should work with you to develop a care plan for this stage of your loved one's care. You will have a say in what treatment and care you want your loved one to have.

At some point you will need to make a choice of whether your loved one stays at home or is transferred to a hospice or hospital.

Copies of your care plan should be distributed to everyone involved but you can change your mind at any stage.

You should be given a list of who to contact 24 hours a day for access to medication and professional advice on symptom control.

You and your family will be under a huge amount of stress and this can affect personal relationships. We have listed useful organisations that can help you and your family come to terms with your loss.

“

**WE PREPARED A MEMORY
BOX, IT HELPED**

”

How can i get more information?

You can get in touch with others in a similar situation through ALD Life who can also provide practical information, support and advice. ALD Life also runs an annual event which brings together families and individuals suffering from all aspects of carrying the ALD gene to learn about innovations in treatment and research and share experiences.

Listed below are just some of the useful organisations that are there to help you:

INFORMATION, SUPPORT AND ADVICE

ALD Life

ALD Life was founded by Sara Hunt after both her sons were diagnosed with ALD. She has first hand experience of most aspects of dealing with the disorder: her elder son, Alex, had symptomatic ALD and was diagnosed at age 7 in 2001, and passed away in 2012. Her younger son, Ayden, had a successful bone marrow transplant in 2008. The charity provides practical, emotional and financial support for all those affected by ALD and AMN.

www.aldlife.org

Tel: 020 7701 4388

Email: info@aldlife.org

Carers Trust

Carers Trust is the largest provider of comprehensive carer support services in the UK. It runs a network of 144 independently managed Carers' Centres and provides quality information, advice and support services to over 400,000 carers.

www.carers.org

Tel: 0844 800 4361

Email: info@carers.org

Carers UK

Carers UK campaigns to make sure carers receive the practical, financial and emotional support they need.

www.carersuk.org

Tel: 0808 808 7777

Email: info@carersuk.org

Citizens Advice Bureau

Free information and advice on legal and money problems and can help you if you experience problems with benefits or housing. Your local branch can be found on the national website.

www.adviceguide.org.uk

Contact a Family

National charity Contact a Family provide advice and support for any family with a disabled child. They run a free helpline to answer all queries on everything from medical advice, to benefits, grants and schooling. They will even send someone to your home to assess what you will need.

www.cafamily.org.uk

Free helpline 0808 808 3555

Email: info@cafamily.org

Directgov

Government website for information about benefits and entitlements

www.direct.gov.uk

Disability Rights UK

Provides advice and support for disabled people on a wide range of issues from claiming the right benefits to employment and independent living.

Ground Floor
CAN Mezzanine
49-51 East Rd
London
N1 6AH

www.disabilityrightsuk.org

Tel: 020 7250 8181

Email: enquiries@disabilityrightsuk.org

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Rainbow Trust Children's Charity

Charity specifically for families who have a child with a terminal or life-threatening illness. Provides practical and emotional support through respite and befriending.

www.rainbowtrust.org.uk

Tel: 01372 363438

Together for short lives

A nationwide charity set up to achieve a better quality of life and care for every life-limited child and their family.

Their website is packed with useful information including a list of UK hospices.

www.togetherforshortlives.org.uk

Tel: 0808 8088 100

EQUIPMENT AND FINANCIAL SUPPORT

Assist UK

Assist UK has a network of local disabled living centres. Each centre includes a permanent exhibition of products and equipment that you can see and try.

www.assist-uk.org

Tel: 0161 850 9757

Email: general.info@assist-uk.org

Dentocare

Provides specialist dental supplies for those with disabilities.

www.dentocare.co.uk

Tel: 0800 980 1517

Directory of Grants for Individuals in Need

Directory of Grants for Individuals in Need available from the reference section of your local library or online at

www.grantsforindividuals.org.uk

Disabled Living Foundation

National charity that can help you find products and suppliers of equipment for disabled people. Also has an equipment demonstration centre.

www.dlf.org.uk

Helpline: 0300 999 0004

Email: helpline@dlf.org.uk

Family Fund

The Family Fund is a charity that helps families with severely disabled children and gives grants for things such as washing machines, driving lessons, costs of hospital visits, computers and holidays.

www.familyfund.org.uk

Tel: 08449 744 099

Email: info@familyfund.org.uk

Motability Scheme

The motability charity helps provide wheelchair adapted cars, powered scooters and powered wheelchairs for disabled people in the UK.

www.motability.co.uk

Tel: 0300 456 4566

Newlife Foundation For Disabled Children

The Newlife Foundation helps disabled and terminally ill children. Provides equipment to help children, nurse-led support services and pioneering medical research.

www.newlifefcharity.co.uk

Tel: 01543 462777

Nurse helpline: 0800 902 0095

The Variety Club

Traditionally the 'showbiz charity', the Variety Club works to help improve the lives of sick, disabled and disadvantaged children and young people up to the age of 19. They donate electric wheelchairs, specialist beds, car seats, sensory equipment, standing frames and many other items that can change the life of a child in need.

www.varietyclub.org.uk

Tel: 020 7428 8100

Email: info@varietyclub.org.uk

Whizz-Kids

Whizz-kids provides disabled children with customised mobility equipment and advice. Also provides activity breaks for those with disabilities.

www.whizzkidz.org.uk

Tel: 020 7233 6600

HOLIDAYS & ONCE IN A LIFETIME EXPERIENCES

Dreams Come True

A national charity that makes dreams a reality for terminally ill children.

www.dreamscometrue.uk.com

Tel: 0800 018 6013

Email: info@dreamscometrue.uk.com

Family Holiday Association

Provides grants towards a one-week holiday of the family's choice. Referral needed from a health visitor or social worker.

www.familyholidayassociation.org.uk

Tel: 020 3117 0650

Email: grantofficer@familyholidayassociation.org.uk

PRACTICAL INFORMATION FOR MALES WITH SYMPTOMATIC ALD

Make-A-Wish Foundation UK

The world-famous Make-A-Wish foundation grants magical dream wishes to children aged 3 –17 who are fighting life-threatening illnesses.

www.make-a-wish.org.uk

Tel: 01276 405 060

Email: info@make-a-wish.org.uk

Starlight Foundation

Another famous charity that makes dreams come true for terminally and seriously ill children.

www.starlight.org.uk

Tel: 020 7262 2881

Email: info@starlight.org.uk

BEREAVEMENT

Childhood Bereavement Network

Provides local and national information, guidance and support for families and carers dealing with the impact of a child's death on their lives.

www.childbereavementnetwork.org

Tel: 020 7843 6309

Email: cbn@ncb.org.uk

The Child Death Helpline

The child death helpline is for anyone affected by the death of a child, however recently or long ago.

www.childdeathhelpline.org

Helpline: 0800 282 986

Email: contact@childdeathhelpline

The Compassionate Friends Helpline

An organisation of bereaved parents who offer understanding, support and encouragement to others following the death of a child. They also offer support to relatives, professionals and friends who are helping the family cope.

www.tcf.org.uk

Helpline: 0845 123 2304

Email: helpline@tcf.org.uk

TCF Sibling Support

TCF Sibling support is run by The Compassionate Friends and provides nationwide self-help for people who have suffered the loss of a brother or sister.

www.tcfsiblingsupport.org.uk

Tel: 0845 123 2304

Email: helpline@tcfsupport.org.uk

Cruse Bereavement Care

A national organisation supporting anyone affected by loss of a loved one.

www.cruse.org.uk

Tel: 0844 477 9400

Email: helpline@cruse.org.uk





INFORMATION SOURCES

The medical information in this leaflet, shaded in red, has been compiled from the following references, and reviewed by an expert doctor.

Engelen M et al. Orphanet Journal of Rare Diseases 2012;7:51.

Ferrer I et al. Brain Pathology 20 (2010) 817–830

Berger J et al. Brain Pathology 20 (2010) 845-856.

The Society for Endocrinology, 2011. Adrenal Insufficiency.

All medical and benefits information is correct at time of going to press January 2015.