Living with A-T

AT society
awareness research care

A-T, short for ataxia-telangiectasia, is such a rare disease that most people don't know about it. It's likely that you heard about it for the first time when you found out that someone you know might have it. It's a progressive disease that causes disability including problems with walking. We understand learning that someone may have A-T is difficult news. There's a great deal of information to take in.

This leaflet gives an overview of what A-T is, how it affects children and adults, and what to expect. There's lots more information on our website about A-T and how the A-T Society can help.

Diagnosis and afterwards

The first signs that a child may have A-T are there when they're very young. When they start walking, for example, they have problems with balance and hold on to things to keep steady. But toddlers are always wobbly, so it isn't always obvious for a while that there's anything wrong.

It can take several months to get a diagnosis of A-T. This is partly because it's so rare, and partly because it has many symptoms, which aren't exactly the same for each person. Some of these symptoms occur in other more common conditions too. There is a blood test that can confirm a diagnosis of A-T. A specialist consultant can do this and the A-T Society can help with a referral.

A-T is usually diagnosed in early childhood and it's usually what's known as 'classic A-T' – there are different types of A-T. A small number of people are diagnosed when symptoms appear later in childhood or as adults. Their disease tends to be less severe and usually it's a type known as 'mild variant A-T'.

Learning that a child or someone in the family has A-T is overwhelming, practically and emotionally. Everything changes. From now on, they'll have many medical appointments with different specialists and other health professionals. A child with A-T will need help at home and at school. It's hard to think about what the future looks like, and life may be very different now from what families imagined or planned.

The A-T Society is here to help. We offer information and support, we can help people get the best medical care and services, and we can help you understand more about A-T and what to expect.

> **I was alone when I received the letter confirming our daughter had A-T. I had no idea what A-T was and went straight onto the internet. Luckily I found the A-T Society website very easily and phoned them. Kay was fantastic and talked to me for over an hour – without the support of the Society that day I really do not know what I would have done.**
>
> *Mother in Cheshire*

Key facts about A-T

A-T is extremely rare: there are only about 200 families in the UK where someone has it. When children have A-T it affects how they develop and function. Adults can be diagnosed too, though this is rarer.

A-T affects both the nervous system and the immune system.
It's a progressive disease meaning that symptoms get worse over time. It's variable meaning that people with A-T don't all have the same symptoms and the disease progresses at different speeds. So while we know it's progressive, it isn't possible to predict exactly what will happen and when for each person.

A-T is a life-shortening disease and at present there's no cure.
Having A-T means that a person has a higher risk of developing cancer. They also have frequent infections which can lead to lung damage. Children diagnosed with classic A-T have lived to their mid-20s on average.

However, it's likely that life expectancy is increasing as we learn more about A-T. In the last 10 to 15 years our understanding and medical management of the symptoms have improved a great deal, so that people can now live longer with a much better quality of life.

What causes A-T? What we know...

A-T is caused by defects in a gene, the ATM gene. About one person in every 250 carries one defective copy of the ATM gene, without knowing it, along with one normal copy. It's only when two people who are 'carriers' have a child together, that their child may have A-T. This happens when the child inherits the defective copy of the ATM gene from each parent. For each pregnancy there is a one in four chance of this happening.

The ATM gene controls how the body produces a particular protein called the ATM protein. This protein repairs cells in the body when their DNA is damaged. People with classic A-T either don't produce this protein at all, or if they do, it doesn't work properly. People with mild variant A-T produce only a small amount of working protein. Therefore when a person has A-T their body can't properly repair damaged cells, and this is what leads to the symptoms.

...and what we don't know, yet

There's a great deal we don't know about A-T. We don't know why it affects some processes in the body but not others: why it affects movement, for example, but not hearing or taste. We don't know why some cells in the body die off without the ATM protein but others don't.

A-T is a difficult disease to research. So very few people are affected that there are problems with collecting enough data to rely on, and it's hard to attract funding. And as well as being rare, A-T is a very complex condition with effects on many of the body's functions.

There is some valuable research currently under way, as researchers interested in treating cancers are looking into the biochemistry of the ATM protein. And the A-T Society has an active research programme, funding studies specific to A-T and co-ordinating work across other countries to help international researchers collaborate. But while we are making good progress, it's likely to take many more years of research to get a full understanding of A-T and to develop effective treatments.

Symptoms

A-T affects both the nervous system and the immune system, and people may have many different symptoms (see page opposite). Not everyone has or will have all of these, and symptoms don't follow the same pattern for everyone. While no treatments can stop the condition progressing, we know much more now about how to manage A-T's symptoms and how to prevent some of the problems. For example, about half of people with A-T have immune problems, and are likely to get repeated lung or chest infections. But treatment with background antibiotics can prevent these recurrent infections, and gives people a much better quality of life. There's more information about symptoms and managing them in our Clinical Guidance document, available on the A-T Society's website.

Children and adults with A-T also have an increased risk of cancer. There's a one in four chance they will develop a cancer in their lifetime, often leukaemia or lymphoma. Treatments may need to be adapted to take account of A-T, but most drugs can be used and cancer can be effectively treated.

People with A-T can't safely have radiotherapy as part of cancer treatment: they are sensitive to radiation because their body can't repair the damage that radiation causes to cells. But they can have X-rays when they're essential for good medical care, for example to diagnose a bone fracture.

People with A-T will have some, but not all, of these symptoms

- Poor co-ordination or ataxia. This usually starts with problems walking as a toddler, and then difficulties with balance and movement as children grow
- Fatigue, an intense tiredness
- Involuntary movements, such as twitches and tremors
- Muscle spasms that cause stiff and twisted positions. This is called dystonia
- Eyes that look bloodshot, with small, dilated blood vessels visible in the whites of the eyes. These are telangiectasia
- Infections of the sinuses and lungs, which over time can lead to lung disease
- Difficulty with eye movements, including following objects or reading
- Drooling. This particularly affects young children when they're tired or concentrating
- Slurred speech, known as dysarthria
- Problems with swallowing
- Slow growth. Many children with A-T are thin and have a small build
- Delayed puberty or incomplete development at puberty
- Premature, age-related changes to their hair and skin

> **Regular monitoring and early proactive treatment of symptoms is crucial in A-T, particularly for those children who have immune problems. This could make a real difference to quality and length of life.**
>
> *Dr Jayesh Bhatt, Nottingham A-T Centre*

Family life with A-T

While children with A-T can and do have full and busy lives, the condition affects them and their families in many ways.

Looking after young children is always demanding, and all the more so when a child has A-T. It's time-consuming to co-ordinate all the medical appointments a child needs, particularly if there are other children in the family to care for too. Many families find that it isn't possible for both parents to continue working full-time, which in turn affects their financial situation. It can be helpful to get advice about benefits and how to claim them. And as a child gets older and starts to make use of a wheelchair, families will also need to think about adaptations at home.

Education is another area to consider. Some children with A-T do well in mainstream primary and secondary schools; some go on to further education; others are in specialist schools. However, they all need support to get the most from school – with communication, for example, if their speech and reading are affected, with getting from place to place around school, with carrying things and at mealtimes.

The A-T Society can give advice about all these matters, to help children and families get the best possible care and support. If and when families want, we can also put them in touch with others who have children with A-T, either online or in person. Many people find it helps to meet others in a similar situation – as a parent, a brother or a sister – and to talk to someone who understands.

"As our son's condition deteriorated it became clear that we needed to adapt our home but we didn't know where to start. With lots of help from the A-T Society and a long fight, we were able to get the adaptations we needed. Now our son has his own en-suite room and is fiercely independent of us!"

Parent of teenage boy near London

Living with changes

Because A-T is a progressive condition, some symptoms will become more severe over time and some new ones will appear.

There will be many changes to adjust to. Most children will start to use a wheelchair, for example, as walking becomes more tiring, and they may be making considerable use of it by the time they're 10.

While it's impossible to predict exactly what will happen and when, we know that A-T tends to progress in uneven steps rather than in a smooth, gradual way. The steps can be very steep and for a while a child's condition can deteriorate rapidly.

This is always concerning for parents to see. But we also know that neurological changes tend to plateau or 'level off' during a child's teenage years: they reach a point and stay there. In this way A-T is different from some other progressive diseases, such as Motor Neurone Disease where the decline inevitably continues. A-T doesn't lead to people living, immobile, with 'locked in syndrome'.

In fact almost all children who have A-T can go on to have some independence as they grow up, and with the right support they can make choices about their lives. Young adults living with A-T, like most young people, say that they value making their own decisions about education, their leisure time and social life.

> **Sometimes people think, 'oh she's disabled so she can't make decisions'. That really gets me cross. I can make my own decisions and I like to be as independent as possible.**
>
> *Young adult with A-T in South-West*

Help and support

The right support and help make all the difference. They are available and the A-T Society is here to help.

A person diagnosed with A-T will see many different doctors and therapists. There are appointments with a neurologist, an immunologist, a respiratory consultant and a genetic specialist. They will probably see an occupational therapist, a physiotherapist, a speech and language therapist, a dietician and perhaps a counsellor.

The best care comes from a partnership between local services, who see people day to day, and specialists at one of our two national A-T centres. Our Clinical Guidance document, available on the website or from our office, gives details of how to organise this.

There are two national centres: one for children in Nottingham and one for adults in Papworth, near Cambridge. The doctors and therapists at these specialist centres have experience of treating many people with A-T, and can give valuable advice to help people keep as well as possible and get the most out of life. We can arrange a referral to one of the centres, and usually people would then visit Nottingham every two years or so, and Papworth about every year. We can put local healthcare professionals in touch with the centres too, for information and expert advice about A-T.

We also help families with getting other services they need. This often means liaising with schools and social services, working with local authorities to help with equipment, helping with applying for benefits and advising on applications and grants for home adaptations.

We can be a listening ear too, always in complete confidence, and we can put you in touch with our counsellor. When times are difficult, emotional support can be just as important as practical help.

We were really pleased to get our daughter, Brooke, seen at the specialist centre very soon after diagnosis. Kay at the A-T Society managed to sort this for us and helped with accommodation. Things were still very raw, but it was good to go and get some answers from the medical team there and to meet a few other people in the same boat as us. We came away understanding A-T better and ways in which we could help to keep Brooke healthy.

Justine and Stefan, Brooke's parents

A-T Society

We're here to support anyone affected by A-T and their family and friends.

Kay Atkins,
Family Support Officer

Tel: 01582 760 733
8.30am to 4.30pm,
Monday to Friday.

kay@atsociety.org.uk
www.atsociety.org.uk

Founded in 1989, the A-T Society is the only organisation in the UK dedicated to providing information and support to people living with ataxia-telangiectasia as well as funding and promoting research to develop treatments and a cure.

Although most of our work is in the UK and Ireland, our information and resources are used by people around the globe and our research work has a world-wide impact.

The A-T Society is entirely dependent on the generosity of its supporters for funding. This booklet is provided for free, but if you are able to, please make a donation to support our work and help ensure we can provide more information and support more people in future. To donate or find out more about our work, please visit **www.atsociety.org.uk**

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