

A-T: THE DISEASE

Ataxia-Telangiectasia (“A-T”) is a rare, neurodegenerative, genetic disease which affects many parts of the body causing severe disability in young children and premature death. The name refers to the resulting poor co-ordination (ataxia) and what appear to be extra visible blood vessels (telangiectasia) which are most noticeable on the whites of the eyes.

A-T is progressive; it affects the cerebellum (the body’s motor control centre) and, in about 70% of cases, also weakens the immune system leading to respiratory disorders. A proportion of children with A-T will develop leukaemia or cancer of the lymph glands.

Defining features are:

- Loss of balance
- Poor muscle co-ordination
- Immune system deficiencies
- Predisposition to cancer

A-T first shows in early childhood, i.e. the toddler stage. The symptoms are lack of balance, slurred speech and perhaps more than a normal number of infections. All children of this age take a little time to develop good walking skills, coherent speech and an effective immune system so it might be some years before A-T is properly diagnosed.

A-T is evident in early childhood

A-T is progressive

A-T is multi-disabling

A-T is, at present, incurable

All children with A-T have a heightened risk of developing cancer, in particular leukaemia and lymphoma.



THE SYMPTOMS

Progressive loss of balance (ataxia)

Prominent blood vessels, especially in the eyes (telangiectasia)

Loss of co-ordination/body tremors

Inability to hold head erect

Slurred speech (dysarthria)

Restricted eye movements

Immune system problems

THE EFFECTS

Probably reliant on a wheelchair by the age of 10 (some even earlier)

None known

Slower reaction time. Cannot easily write, eat, or dress without help

Difficulties in breathing, swallowing and communicating

Speaking becomes a physical effort; conversation can be a slow process

Reading and following moving objects becomes difficult

Frequent chest infections, colds and pneumonia

WHAT CAUSES A-T?



A-T is inherited. It is passed unknowingly by parents to their children. Each of us carries a few faulty genes but in most cases has a compensating healthy gene. If both parents carry the same defective A-T gene, there is a 1 in 4 chance with each pregnancy that the child will be born with A-T. This is ‘recessive inheritance’, and parents of children with A-T are carriers.

Frequency

- A-T is seen in all populations
- It affects males and females equally
- It is a rare disease affecting about 3 people per million in the UK
- Approximately 0.5% of the population are carriers of the mutant A-T gene (‘ATM’)

Prognosis and Treatment

At present there is neither cure nor any treatment that will halt the progressive nature of the disease.

Dr. Elena Boder, the physician and researcher who gave A-T its name, said of the complex nature of the disease:

“A-T is not just a rare disease. It has become the focus of international research because it exhibits features that are of major concern in medicine today: cancer susceptibility, immuno-deficiency, progressive neurological deterioration and premature ageing. It is hoped that A-T may reveal links between them.”

This takes A-T out of the realm of rare, genetic diseases that affect only a small number of families and brings it into the domain of general public health.

DONATIONS



If you would like to make a donation, please make cheques payable to **A-T Society**. Thank you.

I enclose a donation of £.....

Name

Address

.....

.....

Postcode.....

When completed please detach this form and return to:

**THE A-T SOCIETY
IACR-ROTHAMSTED
HARPENDEN
HERTS
AL5 2JQ**

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I have read the notes above and I want all donations I may make to the A-T Society to be treated as Gift Aid donations.

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TO (BANK)
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Please pay to the account of the A-T Society,
HSBC Bank plc, 1 High St, Harpenden, Herts AL5 2RS
Account no: 41248693 Sort code: 40-23-11

The sum of £

Date of first paymentand thereafter each week/month/quarter/year until further notice

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RESEARCH INTO A-T



Recent and future research

Research has led to a better understanding of how A-T progresses and has been able to identify the often unique genetic variations between individual sufferers. However, there is still no treatment. A range of drugs has been tried and will continue to be tried. This systematic screening for candidate drugs will, hopefully, make more likely the prospect of an effective treatment. It is also possible that in the long term an answer may come from new technologies such as stem cell transfer.

ATM and the general population

Defects of the ATM gene (the mutant gene first located in 1995) also affect people other than families with A-T. The ATM gene is reported to be a breast cancer susceptibility gene. In addition, mutations in ATM are seen in chronic leukaemia, the most common form of leukaemia in the UK, and also in other types of leukaemia and lymphoma in the general population.

A-T families and breast cancer

There is evidence of a moderately increased risk of breast cancer in mothers of children with A-T. This risk may be greater in mothers less than 50 years old.

OBJECTIVES



The objective of the A-T Society is to alleviate the distress and suffering that A-T causes.

We do this by:

- supporting children and adults with A-T and their families
- funding research into A-T
- supporting two A-T clinics (Nottingham & Cambridge)
- providing information on living with the disease
- keeping families informed through a newsletter
- raising awareness

Please help us to continue our work by:

- making a donation, no matter how small
- signing a gift aid declaration
- organising a fundraising event
- designating us your company charity
- remembering us in your will

*The A-T Society is a small national charity unique in the services it offers families.
We are grateful for your help and support - thank you.*

For further information, please contact:

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Registered Charity No. 1105528

The Ataxia-Telangiectasia Society WHAT IS A-T?



Supporting the Family - Fighting the Disease