Nutrition in Ataxia Telangiectasia

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- Ataxia telangiectasia is a rare genetic condition that affects movement, motor function, blood vessel dilation and the immune system
- Estimated prevalence of 1:400,000 in the UK[1]
- Nutritional concerns can include dysphagia, malnutrition, aspiration, and obesity
- Incidence of hyperlipidemia, diabetes, vitamin D deficiency is yet unknown
- Paediatric and adult patients have access to specialist care for annual review, and may access local dietetic services throughout their life for nutritional care.

Overview and management

National guidelines on adult care in ataxia telangiectasia are expected to be published in 2016. Paediatric guidelines were published in 2014.[2]

Ataxia telangiectasia (A-T) (also referred to as Louis–Bar syndrome) is a rare, genetically inherited, autosomal recessive neurodegenerative disease causing severe disability. Ataxia refers to poor coordination and telangiectasia to small dilated blood vessels, both of which are hallmarks of the disease.[3] Three different forms of A-T are recognised: classical, variant and A-T like syndrome; often patients with variant disease experience milder symptoms.

The condition affects many parts of the body:

- It impairs certain areas of the brain, including the cerebellum, causing difficulty with movement and coordination
- It may weaken the immune system causing a predisposition to infection
- It prevents repair of broken DNA, increasing risk of cancer.

Diagnosis

Due to the rarity of the condition, children may initially be misdiagnosed with ataxic cerebral palsy, Friedreich's ataxia or Cogan oculomotor apraxia due to a lack of experience with the condition and its symptoms.[4] Diagnosis is confirmed by genetic testing to identify mutations in the ATM genes and assays demonstrating reduced or absent ATM kinase activity. The mode of inheritance for A-T is autosomal recessive. The incidence world-wide is estimated between one in 40,000-100,000 births, with an estimated 1.4- 2% frequency of ATM carriers.[5,6]

Life expectancy

Life expectancy is highly variable, with the mean being approximately 25 years of age,[7] but continues to improve with advances in care. The most common causes of death are cancer, infection and lung failure.[8]

There is no treatment known to slow or stop the progression of the neurological problems. Treatment of A-T is supportive and based upon symptoms. Support from the physiotherapist, occupational therapist, dietitian and speech and language therapist may help maintain function and can have significant improvement on patient, as well as parents'/carers', quality of life, but will not slow the course of neurodegeneration. Certain anti-Parkinson and anti-epileptic drugs may be useful in the management of symptoms, but should be prescribed in consultation with a neurologist.

Features of A-T are quite variable in the severity between different individuals and at different ages. Table one (next page) discusses common features, but these may not be present in all patients.

Nutritional considerations

Similar to the paediatric A-T national guidelines,[2] the main goals remain for feeding and swallowing to be safe and adequate, and for mealtimes to be enjoyable.

With A-T patients, involuntary movements can make feeding difficult or messy and may excessively prolong mealtimes. It may be easier to use finger foods rather than use utensils (e.g. spoon or fork) or speak to the occupation therapist regarding an assessment for adapted cutlery.

Table One: Common Features of Ataxia Telangiectasia

Feature	Symptoms
Ataxia	Difficulty with control of movement that is apparent in the toddler years[9, 10] and worsens in school to pre-teen years. Sometimes the patient will have a problem with standing or sitting still and tends to sway. Around the beginning of their second decade children with typical forms of A-T start using a wheelchair for long distances.
Problems with fine motor function	Includes writing, colouring, and using utensils to eat. Involuntary movements may start at any age and may worsen over time. These extra movements can take many forms, including small jerks of the hands and feet that look like fidgeting (chorea), slower twisting movements of the upper body (athetosis), adoption of stiff and twisted postures (dystonia), occasional uncontrolled jerks (myoclonic jerks), and various rhythmic and non-rhythmic movements with attempts at coordinated action (tremors).
Oculomotor apraxia	Difficulty with coordination of head and eye movement when shifting gaze from one place to the next, e.g. reading.
Telangiectasia	Dilated blood vessels over the white (sclera) of the eyes, making them appear bloodshot.[11]
Abnormalities of the immune system	Occur in about two-thirds of patients.[12] Some people have frequent infections of the upper and lower respiratory tract.
Chronic lung disease	Develops in more than 25% of people with A-T.[13] Three major types of respiratory disease can develop and patients may develop more than one condition: **1.** Recurrent and chronic Sinopulmonary infections. **2.** Lung disease, typically bronchiectasis, caused by ineffective cough, swallowing dysfunction, immunodeficiency and impaired airway clearance **3.** Less commonly interstitial lung disease.
Increased incidence (approximately 25% lifetime risk) of cancers	Increase risk particularly in lymphomas and leukaemia, but other cancers can occur.[14] When possible, treatment should avoid the use of radiotherapy or chemotherapy drugs that work in a similar way, as these are particularly toxic for people with A-T.
Delayed/ incomplete pubertal development	Delayed or incomplete pubertal development and very early menopause.
Drooling	Particularly in young children when tired or concentrating on activities.
Dysarthria	Slurred, slow, or distorted speech sounds.
Premature changes in hair and skin	It can also cause vitiligo (an auto-immune disease causing loss of skin pigment resulting in blotchy 'bleach-splashed' look). Warts can be extensive and resistant to treatment.[15]
Feet deformities	Increases the difficulty of walking due to impaired coordination.
Dysphagia	Common, typically during second decade of life, because of neurological changes that interfere with coordination of mouth and pharynx movements that are needed for safe and efficient swallowing. Coordination problems involving the mouth may make chewing difficult and can prolong mealtimes. Problems involving the pharynx may cause to be aspiration.[16]
Diabetes	In adolescence or later.[16]
Growth retardation	Weight and/or height.[17-19]
Feeding Difficulties	Feeding and swallowing may become difficult for some people with A-T as they get older.[20]
Increased risk for the development of breast cancer	Women, who are A-T carriers, have approximately a two-fold increased risk for the development of breast cancer compared to the general population.[20, 21] This includes all mothers of A-T children and some female relatives. Current consensus is for these women to have earlier and more frequent breast cancer screening.

Feeding requirements vary greatly, from patients only requiring assistance with cutting meat or opening packets in order to self-feed, up to needing full assistance at mealtimes by their parent/carer. Therefore, it is important to monitor the level of assistance that the patient requires as well as time taken to complete the average meal. If the patient attends a day care centre, the staff should be aware that assistance at mealtimes may be needed, and what these specific requirements are.

Dysphagia

The speech and language therapist will provide tailored treatment following assessment for any swallowing concerns. Currently, at the author's centre, the speech and language therapist performs a videofluoroscopy on each new A-T patient at their first annual review and will repeat as per clinical need. Dietitians may be required to support patients with education on following a texture modified diet alongside the speech and language therapist, along with providing support on adapting foods/snacks and/or nutritional supplements to ensure a well-balanced diet as much as realistically possible.

Nutritional support

As with most other patient groups, in practice, a food first approach is promoted and patients are encouraged to have an oral diet as long as they are able. In A-T patients this includes self-feeding for as long as feasible, as the ability to feed themselves can have a big impact on their self-confidence, especially in social situations.

A feeding (gastrostomy) tube is recommended when any of the following occur:[22]

- To supplement nutritional intake partially or fully, to sustain growth (in children) or promote weight gain in underweight patients (BMI under 20 kg/m^2), when all other nutrition support was tried and not tolerated
- Aspiration is deemed to be a high risk and recurrent chest infections are seen

- Mealtimes are deemed to be too stressful or long for the patient, interfering with other activities of daily living. (It is important to assess this from the patient's viewpoint first, and then from the carer/parents point of view to ensure the decision is in the best interest of the patient).

Feeding tubes can decrease the risk of aspiration by enabling the patient to avoid liquids or foods that are difficult to swallow and to provide adequate calories without the stress and time commitment of prolonged meals. Gastrostomy feeding tubes do not prevent patients from eating orally, if they are safe to do so, as this type of feeding can be used alongside oral diet.

At present there is a lack of published evidence on nutrition in A-T due to the rarity of the condition. A study (2011) from the United States of America published data on the safety and caregiver satisfaction with gastrostomy in patients with A-T and the benefits of PEG feeding.[22] This retrospective review of 175 patients who visited the Children's Centre at John Hopkins Hospital suggested that patients may benefit from early discussion and/or placement of PEG feeding. It also showed that PEG feeding in this patient group had a positive impact on meeting their nutritional needs, was associated with easier mealtimes and the carers reported significant improvements in the patient's participation in daily activities.

Vitamin D, antioxidant status and hypercholesterolemia in A-T

A small study[18] showed that 61.5% of A-T children had vitamin D deficiency (< 20 mg/L). This is also echoed in a further study.[23] In view of this, patients' vitamin D levels are now monitored at their annual review at Papworth Hospital to try to establish the prevalence of this issue. As vitamin D deficiency may be a risk amongst this group, care should be taken to ensure adequate vitamin D and calcium intake in the diet.

Although antioxidants have been suggested as a treatment option for reducing oxidative stress, studies have been inconclusive on their value and so should not be actively promoted at high doses, owing to lack of evidence.[19, 24, 25] Antioxidants are naturally found in foods when healthy well-balanced diets are followed. Recent studies have found that A-T patients are likely to struggle with dyslipidaemia[26] and may be offered statins for this. Education on heart-healthy dietary fats is discussed when appropriate but the impact of dietary education on hypercholesterolemia in this disease cohort has not yet been studied. At the Papworth Hospital we have started monitoring lipid profile at annual reviews to assess prevalence.

A few studies suggest that A-T patients may be at risk for diabetes or impaired glucose tolerance, though as yet there is no conclusive data. It is intended that HbA1C monitoring will also be collected at each annual review to assess incidence of this in our population. Education on a diabetic diet is provided if this is diagnosed, and referral completed to a local diabetic clinic.

Obesity in A-T

Obesity is present in a small number of the variant A-T patient population seen at the Papworth Hospital clinic. If not corrected or managed, this could then affect a patient's confidence and mobility, as well as lead to number of lifestyle diseases, such as diabetes or heart disease. Obese A-T patients appear to have low awareness of their current weight as accessing a scale is difficult when a wheelchair user. General weight loss advice should be provided, taking into account the following additional factors:

- Patients may find exercise difficult due to coordination required, so encourage patients to discuss increasing exercise with their physiotherapist.
- Patients may find access to a suitable weighing scale difficult, as most GP surgeries only have standing or chair scales available. Liaising with their local hospital is helpful as they are usually happy to provide patients access to their floor scale to promote monthly self-monitoring of weight.

Dietetic screening and intervention

No observational studies have been performed on nutritional complications and A-T, with few data found on the nutritional implications in adults with this disorder.

The Papworth Adult Ataxia Telangiectasia Service is currently the sole nationally commissioned provider for adults with A-T in the UK, with paediatric services provided by Nottingham Hospital. As patients travel from around the UK, many go on to have interactions with services local to them.

The 'Malnutrition Universal Screening Tool' ('MUST') does not always highlight all nutritional issues in this patient group, so a consensus was reached that, at present, each adult A-T patient is assessed by a dietitian as part of their annual review at the Papworth Hospital, with referral to local dietetic services for follow-up if required. Patients are also reviewed by the multidisciplinary team (including a respiratory physician, neurologist, immunologist, nurse, physiotherapist, speech and language therapist, social worker, occupational therapist) during their annual review inpatient stay. This is especially beneficial if advanced planning around future treatments is needed, such as gastrostomy tubes, or ongoing swallow assessments. This allows patients to absorb the information and ensure they make informed decisions.

Conclusion

The National Adult Guidance on A-T is currently being written and should be available late 2016. The Guidance for A-T Paediatric Care was completed in 2014.[2]

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