ORIGINAL ARTICLE

Nutritional status of patients with ataxia-telangiectasia: A case for early and ongoing nutrition support and intervention

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Aim: Ataxia-telangiectasia (A-T) is a rare genomic syndrome resulting in severe disability. Chronic childhood disorders can profoundly influence growth and development. Nutrition-related issues in A-T are not well described, and there are no nutritional guidelines. This study investigated the nutrition-related characteristics and behaviours of Australian A-T patients attending a national clinic.

Methods: A cross-sectional analysis of 13 A-T patients (nine females; aged: 4–23 years): nutritional status was assessed by anthropometric and body cell mass (BCM) calculations. Parents reported their child's diet history and physical and behavioural factors that affect nutrition including fatigue and need for assistance.

Results: Ten (77%) had short stature (height for age z scores <−1), and seven (54%) were underweight for height (weight/height z scores <−1). Significant malnutrition (BCM z scores <−2) was detected in nine (69%) including the one adult who was severely malnourished. Malnutrition increased significantly with age (BCM for height z scores and age, $r = -0.937$, $P < 0.001$). Eight (62%) patients ate poorly compared with estimated energy requirement for weight. Poor diet quality was characterised by high fat and sugar choices. Parents reported significant nutritional barriers as chronic tiredness and the need for care giver assistance with meals.

Conclusions: This study confirms profound malnutrition in Australian A-T patients. Poor intakes and diet quality suggest the need for early nutrition intervention. Ongoing support for families and early discussions on tube feeding are required to address changing needs in childhood and likely nutritional decline into adulthood. A prospective study is required to assess feasibility and effectiveness of nutrition interventions in young people with A-T.

Key words: developmental; metabolic; neurology; nutrition.

What is already known on this topic

- 1 A-T is a rare disorder resulting in severe disability.
- 2 Chronic childhood disorders can profoundly influence nutritional status, growth and development.

What this paper adds

- 1 Describes profound malnutrition in Australian A-T patients.
- 2 Exposes poor intakes and diet quality as targets for intervention, and chronic fatigue and need for assistance with meals as major contributors.
- 3 Highlights the need for nutrition intervention and ongoing support in chronic disease from an early age.

Ataxia-telangiectasia (A-T) is a rare genomic instability syndrome caused by a defective DNA damage response characterised by systematic inflammation and neurological impairments.^{1,2} Multiple clinical pathologies appear from 12 months of age and

can include neuro-degeneration and poor motor co-ordination, a weakened immune system, and reduced life-span.^{3,4} Chronic childhood diseases have a profound influence on growth and development,^{5–7} and patients with better growth and nutritional status have better prognosis and increased survival.⁸

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In A-T, failure to thrive has been reported in 58% and 72% of A-T patients in early British studies^{9,10} and more recently in 38% of Iranian A-T children.¹¹ Schubert *et al.* reported low BMI in 84% of A-T subjects (aged 4–24 years),¹² and Kieslich *et al.* reported low BMI growth status in 55% of A-T subjects (aged 8–26 years).¹³ Causes may be due to a number of factors including oropharyngeal dysphagia and aspiration.^{14,15} However, oxidative stress, inflammation and silent lung disease may also

contribute. A study of 27 A-T subjects reported 41% had obstructive lung disease.¹⁶ A larger study of 70 A-T subjects found oropharyngeal dysphagia in 73% including 14 described as aspirators.¹⁴ Aspirators were older and had lower bodyweight and weight for height *z* scores (WHZ). An analysis of survival probability suggests premature death is the result of secondary complications rather than an inevitable consequence of A-T.⁴

The literature suggests A-T patients are vulnerable to issues that influence their nutritional status. Adequate early nutrition intervention and support may help patients attain better growth and improve prognosis.¹⁵ However, there are no established guidelines for nutritional assessment and management in A-T. The aim of the current study was to investigate the nutritional status, dietary intakes, and nutrition-related characteristics and behaviours of Australian A-T patients. The results will inform their nutritional management.

Methods

This is a cross-sectional analysis of data collected from patients who presented in 2012 to a national A-T dedicated clinic (neurologist-led with occupational therapy, physiotherapy, orthotics, respiratory, immunology, audiology, dietetics, genetic counselling). The actual number of Australians with A-T is unknown, but the 23 A-T patients registered with the clinic at the time of the study were from across five Australian states and one territory and were considered representative of the Australian A-T population (based on world-wide population prevalence data of one in 88 000 live births).

All patients had been clinically diagnosed with A-T in accordance with World Health Organisation (WHO) recommendations.¹⁷ A convenience sample of patients was approached consecutively for consent. Patients receiving 90% or more of their nutrition via a gastrostomy tube were excluded. The parents of all participants provided informed written consent. Patients 16 years or older also provided consent. Ethical approval was provided by the Hospitals' Human Research Ethics Committee, and the trial was publicly registered (Australian New Zealand Clinical Trials Registry ACTRN12612000207819).

Height and bodyweight were measured using standard scales correct to 0.1 kg. Each child was laid in a horizontal position and passed through a whole body counter for measuring total body potassium (TBK). Potassium-40, a naturally occurring isotope found in the active cells of the body, is the gold standard for predicting body cell mass (BCM), the total mass of all metabolically active tissue. BCM depletion is a marker of body wasting and therefore nutritional status^{18,19} and has been associated with adverse outcomes.^{20,21} A dietitian conducted interviews with parents, including a diet history using a validated format and food frequency checklist²² and the simplified nutritional appetite questionnaire (SNAQ),²³ a validated four-item multiple choice questionnaire to predict weight loss (≤ 14 , significant risk of $\geq 5\%$ weight loss within 6 months). Parents were asked about need for assistance at meal times as well as a nine-item questionnaire on their child's tiredness and fatigue. All questionnaires were completed immediately or taken home and returned in a reply paid envelope.

Body mass index (BMI) was calculated to assess weight for height using WHO criteria.²⁴ For those under 20 years, per-

tiles for height were compared with growth charts with reference to age and gender.²⁵ *z* Scores (standard deviation (SD) scores) were determined in individuals 5–19 years for BMI for age (BMIZ), height for age (HAZ) and WHZ.²⁶ BCM for height *z* scores (BCM_{HZ}) were also calculated. Individual's resting energy expenditure (REE) was measured by indirect calorimetry using a metabolic cart and hood apparatus. Schofield height and weight equations were used to predict basal metabolic rate (BMR).²⁷ Recommended dietary intake data were used to estimate energy requirement (EER) for gender with 1.6 factor for light physical activity level.²⁸

Food intake data were converted to megajoules (MJ) by entering into FOODWORKS PROFESSIONAL (V.7.0.3016 Xyris Software, Brisbane, Australia, 2012) with AUSNUT food composition database. Average daily intakes were compared with requirement (EER). Individual food items were categorised into the five foundation (core) food groups and discretionary (non-core) food choices described in the Australian Dietary Guidelines.²⁹ The intakes from each food group were reported as a proportion of total energy consumed.

Data were entered into SPSS for Windows 26 (Version 20.0, 2011, IBM Corp., Chicago, IL, USA). Quality assessment of *z* scores used WHO cut-offs for data exclusion:²⁰ -6 and $+6$ HAZ; -5 and $+5$ WHZ. Data were tabulated as frequency (percentage) and mean and standard deviation. Inter-class correlation analysis of relationships between measures used Pearson two-sided coefficient with significance, $P < 0.05$.

Results

Of the 17 patients who attended the clinic, three declined to participate, and one was ineligible due to receiving 90% of nutrition via a gastrostomy tube. Consent was obtained from 13 patients including one adult. Participants were from Western Australia ($n = 1$), Victoria ($n = 5$), New South Wales ($n = 4$) and Queensland ($n = 3$).

Table 1 outlines individual participant characteristics. Compared with population data, three (23%) children had short stature (below third percentile height for age). HAZ indicated that nine children were under the reference height for age: five marginally stunted/mildly malnourished ($-1SD$), three moderately stunted/malnourished ($-2SD$) and one severely stunted/malnourished ($-3SD$). WHZ indicated six were underweight for height: four marginally wasted, one moderately wasted and one severely wasted. The one adult was identified as severely underweight/malnourished using traditional BMI.

BMIZ identified just one child as malnourished ($-3SD$) but identified three children as being overweight or obese ($>+1SD$ and $>+2SD$, respectively). According to BCM_{HZ}, however, nine (69%) of all participants were malnourished ($-2SD$ below the reference value). HAZ was the measure most strongly correlated with BCM_{HZ} ($r = -0.588$, $P = 0.055$), and age was a significant determinant of increasing malnutrition (BCM_{HZ} and age, $r = -0.937$, $P < 0.001$).

Figure 1 shows the energy intakes (MJ/day) of individuals compared with predicted BMR, EER and measured REE. The intakes of two participants included supplementary overnight feeds (via naso-gastric tube and percutaneous gastrostomy tube,

Table 1 Baseline age, gender, anthropometry and z scores of individuals with ataxia telangiectasia (A-T) (presented in ascending order of age, $n = 13$)

Study no.	Age (years)	Gender M/F	Height (cm)	Body weight (kg)	BMI (kg/m ²)	BMIZ (SD)	HAZ (SD)	WHZ (SD)	BCMZH (SD)
11	3.9	F	101.7	16.8	16.2	–	–	–	–
1	7.0	M	113.0	18.8	14.7	–0.5	–3.0	0.0	–1.4
10	7.2	F	111.6†	20.9	16.8	+0.5	–2.0	0.0	–2.3
5	7.3	F	118.3	19.6	14.0	–1.0	–1.0	–1.0	–1.7
8	8.1	F	119.4	29.6	20.8	+2.0	–1.0	+2.0	–3.2
3	8.7	M	120.4†	20.1	13.9	–1.5	–2.0	–1.0	–3.7
9	8.8	F	142.2	48.9	24.2	+2.5	+1.0	+2.0	–1.9
4	9.8	M	130.0	24.5	14.5	–1.0	–1.0	–1.0	–3.2
6	13.1	F	146.0	37.5	17.6	–0.5	–1.0	–1.0	–5.1
2	15.0	F	163.0	36.7	13.8	–3.0	0.0	–3.0	–5.1
13	15.1	M	163.5	47.2	17.6	–1.0	–1.0	–2.0	–4.3
12	15.3	F	149.0†	67.5	30.4	+2.0	–2.0	+2.0	–4.5
7	22.6	M	196.7	62.0	16.0‡	–	–	–	–7.9

†Short stature <3rd percentile for age²⁶. ‡Adult BMI < 18 kg/m² (severely underweight/malnourished).²⁵ The following were applied to individuals aged 5–19 years: –1SD < HAZ or WHZ < 0 well nourished; –2SD < HAZ or WHZ < –1SD mildly malnourished; –3SD < HAZ or WHZ < –2SD moderately malnourished; WAZ < –3SD severely malnourished.²⁷ BMIZ: <–2SD thinness; <–3SD severe thinness; >+1SD overweight equivalent to BMI 25 kg/m² at 19 years; >+2SD obesity equivalent to BMI 30 kg/m² at 19 years.²⁷ BCMZH <–2SD malnourished¹⁹.

–, not calculated; BCMHT, body cell mass for height; BMI, body mass index; F, female; HAZ, height for age; M, male; SD, standard deviation; WHZ, weight for height.

Fig. 1 Energy intake compared with estimated energy requirement for measured weight of individuals with ataxia-telangiectasia (A-T) (presented from left to right in order of age, $n = 13$). *Supplementary overnight tube feeding included. ■ BMR; ■ REE; ■ EER; □ intake.

respectively). REE was expected to approximate 110% of predicted BMR but varied from 141% to 97% across individuals. Only five of the 13 participants (the three youngest and the two receiving tube feeds) met or exceeded their individual EER. Eight participants consumed substantially less energy for their bodyweight requirements, including the three children identified as overweight or obese. Strong correlations were found between REE and BCM data ($r = 0.966$, $P = 0.000$), BCMHZ (0.698, $P = 0.007$), and REE and height ($r = 0.928$, $P = 0.000$).

Figure 2 shows mean intakes from food groups as a proportion of total energy. Discretionary food choices contributed the most energy, consisting mainly of items with high added sugar and fat. Of the five foundation food groups, grain (cereal) foods contributed the most energy. Meat contributed the next largest food group portion, mainly lean meats and small amounts of fish and eggs. Only two children reported eating nuts and seeds. Milk products were well represented, mainly whole milk. Vegetable and fruit consumption were relatively low. Supplementary feeds provided 56% and 44% of energy, respectively, for

Fig. 2 Proportional intakes (% energy) of food items from the five foundation (core) food groups and from discretionary food choices consumed by young people with ataxia-telangiectasia (A-T) ($n = 13$).

the two children with tubes but only provided 8% of the total intake of the study group.

The appetite questionnaire ($n = 12$) indicated just two children with poor appetite (SNAQ ≤ 14). However, parents indicated their children were 'fussy eaters', ate a narrow range of foods and commonly experienced taste fatigue. Some parents also reported their children were wary of 'troublesome' foods after incidents of choking, and some reported extended and/or frequent airway infections that impacted their child's appetite (seven children reported at least one airway infection in the 3 months prior to the study). A strong correlation was found between BCMHZ and the SNAQ score ($r = 0.629$, $P = 0.019$).

Table 2 outlines patterns of tiredness and fatigue ($n = 12$), described by all families as substantial and progressive and impacting daily activities including the need for increased assistance. Parents reported encouraging their child to rest in the afternoons to avoid being too tired to eat dinner.

Table 3 outlines the need for assistance with meals ($n = 13$). Most children needed their food cut up into bite-sized portions due to poor motor control and chewing fatigue, and eight individuals used a straw and/or sipper cup to reduce spillage and overcome swallowing difficulties. Even independent eaters needed extra assistance at the evening meal due to tiredness and slowness to feed.

Table 2 Number of young people with ataxia-telangiectasi (A-T) who experience tiredness and fatigue

$n = 12$	Parental agreement	
	Female $n = 8$	Male $n = 4$
Tiredness and fatigue		
Child usually feels energetic in the mornings	8	4
Tiredness/fatigue progress throughout the day	8	4
Child unexpectedly wants to nap during the day	5	2
Fatigue can come on gradually or abruptly	4	2
Fatigue is a daily occurrence	4	1
The following make the tiredness/ fatigue worse		
School days	3	1
Extra activity	3	1
Fatigue is equally mental and physical	5	2
At least three aspects of your child's life are affected by fatigue (e.g. school, homework, social, play, emotional, eating)	8	4
The fatigue has worsened over the past 3 months	3	0

Discussion

This is the first assessment of nutritional status in Australian A-T patients. It demonstrates that those with A-T are vulnerable to issues that influence their nutritional status. These can be overlooked where there are multiple clinical pathologies to address. The contributors to inadequate nutrition were both physical and

behavioural and pose challenges for carers and professionals alike.

Compared with a group of 226 children with cystic fibrosis (CF), the prevalence of short stature in our A-T children was high (23% A-T vs. 14% CF). Critically, the majority (54%) of

Table 3 Number of young people with ataxia-telangiectasia (A-T) needing assistance at meal times

Need for assistance at meal times	4–10 years <i>n</i> = 8	Over 10 years <i>n</i> = 5
Needs assistance with feeding	7	1
Regularly uses a straw/sipper cup	6	2
Uses modified cutlery/plate	2	1
Overnight tube feed to supplement oral intake	0	2

A-T patients were underweight even when their shorter stature was accounted for using WHZ. Where the calculation of BMI was useful for determining adult malnutrition, BMI *z* scores for children were not. Alternatively, BCM depletion was a more accurate representation of nutritional status and found the prevalence of malnutrition to be high (69% A-T vs. 33% CF³⁰) and supports the findings of earlier studies in A-T using conventional methodology.^{7,9,10,12} Of particular note, three children identified as overweight or obese using standard anthropometry were identified as malnourished (*n* = 2) or moderately BCM depleted (*n* = 1). Significantly, BCM depletion worsened with age (*P* < 0.001) where all participants over 9 years of age were identified as malnourished and the one adult participant was severely malnourished. While TBK is the gold standard for estimating BCM, others may not have access. HAZ had the strongest correlation, and bioelectrical impedance analysis may be a useful surrogate in clinical circumstances.³¹

Poor energy intakes were demonstrated for the majority of participants with the exception of the very youngest and those on supplementary tube feeds. When energy requirements were adjusted for actual bodyweight rather than age, intakes remained sub-optimal. However, measured REE demonstrated wide variation when compared with predicted BMR for individuals but was strongly related to BCM data as a more accurate measure of metabolism. Most energy intake came from discretionary food choices, mainly energy dense, high-sugar/high-fat food items. Parents reported 'fussy' eating and avoidance of foods that may cause choking, as well as taste fatigue and increased need for carer assistance especially at the evening meal. Parental factors such as low socio-economic status and educational status were not assessed. However, the barriers to adequate nutrition likely reflect similar issues confronting any parent with a chronically ill child. Where the level of assistance might be anticipated to increase with age and increasing disability, the parents of older children reported the least need for assistance. This might be explained by observations that the youngest appeared very dependent and less able to regulate their activities to conserve energy. In contrast, the families of older children described greater routine and adaptation around their child's energy levels and to support their independence. False perceptions by parents around the level of independence of their child should also be considered.

These Australian findings support early international studies reporting failure to thrive^{9–11} and a high prevalence of low

BMI^{12,29} in young people with A-T. Studies by Schubert *et al.*²⁹ and Voss *et al.*³² also reported low levels of growth hormone. However, our study demonstrated dietary, physical and behavioural factors that might be modified to improve nutritional status. The results also support the findings of Lefton-Greif *et al.* where dysphagia and aspiration appear to be common issues,¹⁴ perhaps resulting in avoidance of 'problematic' foods and increased need for assistance. Early consideration of supplementary gastrostomy feeds may be warranted.¹⁴ Our study is the first to report on tiredness and fatigue in A-T as a major barrier to adequate nutrition, reported by all families. A search of the literature found only one mention of fatigue in relation to A-T, a single case study of an adult with A-T with very high white blood cell count which markedly improved after treatment.³³

While our study reports small numbers (*n* = 13), they represent 57% of the total of 23 patients registered with the Australian clinic in 2012. The possibility that those who did not attend were too sick to travel or felt their needs were being met locally is a possible source of bias. The lack of a comparison group was addressed through comparison with reference measures obtained from healthy childhood populations. The data were also compared with known benchmarks including BCM. All measurements were conducted by a skilled technician to ensure accuracy. The limitations related to collecting dietary data are well known and include recall bias. In most cases, both parents were interviewed to provide increased confidence in reporting. While parents of the three overweight or obese children in the study may have underreported intakes, these children's BCM values were depleted with the two overweight participants identified as malnourished. While the appetite questionnaire did not suggest appetite was a problem in this group, a strong correlation between appetite and BCMHZ scores suggests a relationship between appetite and declining nutritional status. The appetite questionnaire (SNAQ) was completed by parents who may have accepted a lower level of appetite as 'normal' for their A-T child and/or underestimated the impact of frequent fatigue and illness.

This is the first study to confirm profound malnutrition in Australian A-T patients, including in those reported as overweight using conventional anthropometric methods. The study also indicates the progressive nature of malnutrition with age and is the first study to adequately describe poor oral intake, diet quality, and chronic fatigue and the need for care giver assistance with meals. The results demonstrate there is a need for early nutrition intervention in childhood chronic disease and ongoing nutrition support for families including early discussions on tube feeding.¹⁴ Prospective trials are required to assess the feasibility and effectiveness of nutrition interventions in young people with A-T.

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