

Table 1 Patients characteristics before HSCT

Patient No.	Age at HSCT (years)	Deoxyuridine/Thymidine level at diagnosis (normal <0.05 mmol/L)	Genetic mutation <i>TYMP</i>	Weight at HSCT (Kg)(SD per age)/ BMI(SD per age)	Gastrostomy before HSCT (time of placement prior to HSCT)	Bowel obstruction and surgical intervention	TPN	GI symptoms *	Demyelinating polyneuropathy (motor/ sensor)/ White matter changes on brain MRI
1.	10	10.3/Not done	c.1211insT homozygous	23.8 (-3SD)/ No data	No	No	Yes	Mild	Yes/Yes
2.	16	7.6/3.4	c.1160-1G>A homozygous	28.4 (-4SD)/ No data	Yes (1 year)	No	Yes	Moderate	Yes/Yes
3.	27	12.1/6.7	c.1160-1G>A homozygous	30 (-4SD)/ No data	Yes (1 year)	Yes	Yes	Severe	Yes/No data
4.	20	12.2/7.3	c.1160-1G>A homozygous	32.6 (-4SD)/ No data	Yes (1 month)	No	No	Mild	Yes/Yes
5.	10	7.9/10.5	c.433G>A homozygous	28.5 (-1SD)/ 13.6 (-2SD)	No	No	No	Mild	Yes/No data
6.	8	18.6/17.3	c.433G>A homozygous	16.7 (-2SD)/ 12 (-3SD)	Yes (1 month)	No	No	Severe†	Yes/No data

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Abbreviations: HSCT, hematopoietic stem cell transplantation; SD, standard deviation; BMI, body mass index; TPN, total parenteral nutrition; GI, gastrointestinal; MRI, Magnetic resonance imaging

*GI symptoms: Borborygimi, abdominal pain, nausea, vomiting, gastric dilatation (radiological GI signs or by gastroscopy), abdominal distension, dysphagia, constipation, diarrhea, peritonitis, pseudoobstruction, obstruction, weight loss, thinness, cachexia, pancreatitis

Mild GI symptoms: no daily GI symptoms, no need for gastrostomy; TPN was recommended for improving nutritional status

Moderate GI symptoms: daily GI symptoms with gastrostomy required for stomach drainage and TPN as main nutritional source

Severe GI symptoms: daily GI symptoms requiring TPN as main nutritional source and gastrostomy and/or bowel obstruction with surgical intervention

†Clinical status pre-HSCT was severe enough to commence TPN as main nutritional source and placing gastrostomy (daily symptoms and severe episodes of bowel obstruction), however due to limited medical local services those interventions did not take place