Treacher Collins syndrome

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Background information

The patient, a four-month old female (corrected age 3 months), was born at 36 weeks of gestation on 30 May 2011 via emergency Caesarean section, due to pre-eclampsia toxaemia to a 47-year-old mother. She is the fourth child (gravida 4, para 4) of nonconsanguineous parents. She was admitted to hospital on 14 September with difficulty breathing, and was subsequently diagnosed with Treacher Collins syndrome (TCS) and failure to thrive.

Patient's hospital course

During an interview, the mother reported that since birth, the infant has struggled to feed and snored significantly. Through a sleep study, it was noted that the infant suffered frequent and repeated episodes of desaturation. She was subsequently diagnosed with obstructive sleep apnoea due to upper airway obstruction, and a tracheostomy was recommended. On 29 September, the mother reported that milk came out through the patient's nose while she was feeding orally, and she was presumed to have gastro-oesophageal reflux disease (GORD) that was to be diagnostically confirmed. On 3 October, she underwent surgery for the tracheostomy placement. The patient recovered well, and experienced one more episode of nasal regurgitation on 6 October, whilst being fed orally. The nasal regurgitation resolved spontaneously post-tracheostomy placement.

Her weight improved post-surgery, and once she was able to complete all her feeds orally. Her parents were trained on tracheostomy care. The patient was discharged with a nutritional therapeutic programme (NTP) referral, to ensure catch-up growth.

Diagnosis

The diagnosis was TCS with variable expression, upper airway obstruction, obstructive sleep apnoea, and failure to thrive.

Anthropometry

The patient's anthropometric data (Table I) indicates that at birth, it was appropriate for gestational age, and that no intrauterine growth retardation was present. Her birthweight fell on the tenth percentile, and her length and head circumference were on the 50th percentile on the Babson and Fenton growth chart. However, she only gained a total of 560 g during the 14 weeks following birth, and prior to her hospital admission. This was considered to be inadequate since she only gained an average of 5 g per day, or 51 g/week, which is half of the expected weight gain for an infant.[1]

Her nutritional status changed from underweight and mildly wasted (acute malnutrition) at birth, to severely underweight with acute-on-chronic malnutrition. During her admission to the paediatric hospital, her anthropometric status showed a trend of improvement. She achieved an average daily weight gain of 20-30 g/day, which is adequate for catch-up growth.[1]

Diet history at home

The mother reported that the infant struggled to feed since birth, and never successfully latched on to the breast. The mother then attempted bottle-feeding, but the infant tired easily, and only finished three bottles a day, which made up a total volume of 375 ml of Nan 1® per day. This provided her with 251 kcal (90 kcal/kg), 5.6g (2g/kg) protein, and 375 ml (134ml/kg) of fluid, less than the daily recommended values for an infant of her age.[1]

Nutritional management

TCS, also known as Mandibulofacial dysostosis and Franceschetti-Zwalhen-Klein syndrome, is a rare autosomal dominant disease with variable expressivity, which is thought to occur in 1:25 000-50 000 live births.[2-4] This innate disorder is exemplified by many developmental irregularities, that are restricted to the head and neck.[3] The syndrome, which commonly appears with Pierre Robin

Table I: Anthropometric data

Date	Weight (g)	% EWA[a]	Length (cm)	% EHA[b]	% EWH[c]	Nutritional status classification	HC[d] (cm)
30 May 2011 (Birth)	2 260	81%	47	100	81	Underweight and mildly wasted	33
22 September 2011 (Admission to hospital)	2 820	41%	-	-	-	Severely underweight	-
26 September 2011	2 770	49%	-	-	-	Severely underweight	-
29 September 2011	3 000	54%	-	-	-	Severely underweight	-
4 October 2011 (Day 1 post-surgery)	2 800	50%	-	-	-	Severely underweight	-
11 October 2011	3 110	56%	54	91	71	Severely underweight, mildly stunted, and moderately wasted. Acute-on-chronic malnutrition	-
20 October 2011 (Discharge)	3 375	60%	56	94	77	Severely underweight, mildly stunted, and moderately wasted. Acute-on-chronic malnutrition	-

a = percentage of expected weight for age, b = percentage expected height for age; c = percentage expected weight for height, d = head circumference

Sequence, previously known as Pierre Robin syndrome, is caused by mutations in the TCOF1 gene, which is located at chromosome 5q32-q33.1.[2-5] Sixty per cent of cases are due to spontaneous mutations, and forty per cent are familial.[3] Presentation of the condition varies from unnoticeable to severe.[2,3] Diagnosis of the condition is made based on clinical signs and symptoms, which include micrognathia (a small jaw and chin); hypoplastic facial bones; a high palate; anti-Mongoloid eyes; coloboma (a structural defect of the eye, especially in the choroid, retina, or iris), absent, small, or unusually developed ears; and in about 35% of cases, a cleft palate.[2-5] In severe cases, underdevelopment of the facial bones may restrict the infant's airways, resulting in life-threatening respiratory problems.[2-5] Upper airway obstruction (UAO) should be suspected when an infant experiences feeding difficulties resulting in inadequate weight gain, which can be insidious.[6] GORD is often also present with UAO, due to the negative pressure that is created by increased respiratory effort against a partial upper airway obstruction.[6] The medical procedures frequently used to alleviate respiratory distress in these infants include a tracheostomy or mandibular distraction osteogenesis.[2-5]

Micrognathia (mandibular hypoplasia) causes the upper and lower jaw to be improperly aligned, and the receding chin displaces the tongue posteriorly. This results in the infant having difficulty with compressing the feeding nipple and obtaining a seal, which results in a less efficient suckling pattern.[6] Depending on the extent of anomaly present, infants with TCS may experience difficulty with effective swallowing of liquids, and the attainment of adequate nutrition.[2] In cases where the infant is unable to achieve adequate nutritional intake and hydration, tube feeding is initiated, and in severe instances a gastrostomy tube is placed.[4]

The patient presented with the following features: anti-Mongoloid eyes, malar hypoplasia, micrognathia, absence of lower eyelashes, a coloboma, malformed ears, external ear canal deficit, hair on the lateral part of the cheeks, airway-related abnormalities, a high palate and incoordinate swallowing. She did not present with a cleft palate, which is sometimes present in TCS. Neither of her three older siblings, nor her parents, showed any signs of TCS.

On admission to the country hospital, her weight was 2.8kg. She was fed expressed breastmilk with added breastmilk fortifier (FM85) via a nasogastric tube, but her weight remained unchanged at 2.8kg.

On admission to the paediatric hospital, the infant's nutritional management was initiated according to the catch-up growth requirements as prescribed by the World Health Organization (WHO).[7] She was started on five bolus feeds of 65 ml each, of a nutrient-dense feed Infatrini® (Nutricia), via nasogastric tube (NGT). This provided her with 325 kcal (142.5 kcal/kg), 8.45g (3.7g/kg) protein, and 142.5ml/kg fluid. Her feeds were then changed to three oral feeds of 45ml each, and four bolus feeds of 45 ml each via NGT. She tolerated the oral feeds fairly well. On 26 September, her weight had dropped to 2.77 kg, and her feeds were increased to eight feeds of 50 ml each via NGT, in order to promote catch-up growth. On 27 September, it was once again noted that she tired quickly while being fed, and the mother reported that the infant experienced nasal regurgitation when fed orally. By 29 September, her weight had increased to 3.02 kg, and she was receiving five feeds of 70 ml each via NGT. From 30 September-2 October, she had an inadequate intake, due to being kept *nil per os* in anticipation of surgery, and her weight dropped to 2.8 kg. She underwent surgery on 3 October 2011.

On 4 October, day one post-surgery for tracheostomy placement, she was restarted on Infatrini® at 14 ml/hour nasogastrically, which provided her with 336 kcal (120 kcal/kg) and 8.7g (3.12g/kg) protein. By 6 October, she was taking half of her feeds orally, and

only one episode of nasal regurgitation was noted. On 7 October, her weight was 3.03 kg, and she was receiving six oral feeds of 45 ml each, and a top-up feed of 60 ml via NGT. On 11 October, her weight had increased to 3.11 kg, and she was taking all her feeds orally without any nasal regurgitation. By this stage, she was receiving six feeds of 75 ml each of Infatrini®, which provided her with 450 kcal (148.5 kcal/kg), 11.7 g (2.86 g/kg) protein and 450 ml (148.5ml/kg) fluid. She was medically stable, and consequently discharged. On discharge, she was referred to the NTP for PreNan® (Nestlé) with dietary requirements of 150 kcal/kg and 4 g/kg protein for catch-up growth.

Discussion

TCS needs to be managed by a multi-disciplinary team, which includes a plastic surgeon, audiologist, a speech and language therapist (SLT) to alleviate feeding difficulties, and a dietitian, to ensure the infant's nutritional needs are being met.[2-5,8] Feeding difficulties and failure to thrive is common among these infants due to the presence of UAO, and these patients may initially have to be fed via the nasogastric route.[7,8] Only once the UAO has been addressed, can oral feeding be introduced, whilst nasogastric feeding continues until such a stage where the infant is able to meet all his or her nutritional requirements via oral feeding.[9,10] In order not to tire the infant, it has been recommended that initially, oral feeding attempts should be of a short duration (15-20 minutes, or even less), and that other feeding facilitation methods, as recommended by the SLT, are used.[8]

As the infant grows and the face develops, the infant's airway status naturally improves, and generally those who had a tracheostomy can be decannulated.[9,10] Their nutritional prognosis is usually good, since their oral intake improves following either the creation of a tracheostomy, or mandibular distraction osteogenesis.[9,10] These children go on to develop normally. They have normal intelligence, and their feeding and developmental milestones are similar to those of a normal child.[8] Once they have undergone plastic surgery when they are older, TCS sufferers are able to live a good quality life.[8]

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NSSA newsbits

Prof Philip Calder, a professor in nutritional immunology, is the international invited speaker of the Nutrition Society of South Africa. His research focuses on the influence of dietary fatty acids on aspects of cell function and human health, in particular in relation to cardiovascular disease, inflammation and immunity. A range of anti-inflammatory and immunomodulatory effects of the n-3 family of polyunsaturated fatty acids, particularly those found in fish oils, have been identified. The findings of this research aid our knowledge and understanding of the health benefits of naturally occurring fatty acids, and provide information to underpin the design of nutritionally based therapies for use in situations where components of the immune and inflammatory systems are under- or overactive. Some of his current projects investigate the effect of increased intake of salmon by pregnant women on nutrient status and markers of immunity in newborn infants and their subsequent risk of allergic disease, and the effect of marine n-3 fatty acids on inflammatory markers in lean and obese humans.

Prof Calder is well known as a lively presenter, and has the ability to explain highly scientific principles and point out how to translate the information into practice. He has received numerous awards, including the Sir David Cuthbertson Medal awarded by the UK Nutrition Society, the Belgian Danone Institute Chair, the Nutricia International Award, and the New Zealand Nutrition Society's Muriel Bell Award. Prof Calder is a visiting professor at King Saud University in Riyadh, Saudi Arabia. His publications are also cited by a large number of international scientists in high-impact journals. Editor-in-Chief of British Journal of Nutrition and serves on the editorial boards of various other international nutrition journals.