Journal of Paediatrics and Child Health

doi:10.1111/jpc.15434

ORIGINAL ARTICLE

New Feeding Assessment Scale for individuals with genetic syndromes: Validity and reliability in the CHARGE syndrome population

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Aim: To develop a feeding scale for parents/care givers of individuals of all ages with genetic syndromes experiencing extensive feeding and swallowing problems. Second, to assess its validity and reliability in CHARGE syndrome.

Methods: The new Feeding Assessment Scale (FAS) was adapted from a scale for children who need prolonged tube feeding (Paediatric Assessment Scale for Severe Feeding Problems, PASSFP). Ten parents piloted the new scale before it was sent out with the PASSFP and feeding history questions. A subset completed the new scale again 4–8 weeks later.

Results: One hundred parents of individuals with CHARGE syndrome participated from around the world. The new scale had good construct validity, with a significant effect for an increased number of feeding risk factors having higher scale scores ($P < 0.001$). Face validity was high, as scores significantly differed between individuals whose parents identified their feeding difficulties as very mild, mild, moderate, severe and very severe ($P < 0.001$). Test–retest reliability ($r = 0.94$, $P < 0.001$) and internal consistency (Cronbach's alpha 0.91) were both high. There was significant convergent validity between the new scale and the PASSFP ($r = -0.79$, $P < 0.001$).

Conclusions: This new tool is reliable and valid for parents/care givers of individuals with CHARGE syndrome. It can be used to assess the current severity of feeding difficulties and to track progress before and after treatment. It expands upon previous existing tools in that it can be used in both individuals who are not tube fed, as well as in those who are, as well as across the life-span.

Key words: general paediatrics; genetics; gastroenterology; rehabilitation; behavioural.

What is already known on this topic

1 Children, adolescents and adults with genetic syndromes or neurological dysfunction are at high risk for feeding and swallowing issues.
2 Existing tools to assess feeding and swallowing difficulties in this type of patient population are often limited to those who are tube fed, and do not often include those with more mild difficulties.
3 Existing tools are also often limited to either childhood or adulthood, and do not encompass the entire life spectrum.

What this paper adds

1 Our new feeding scale is designed for use across the life-span, from infancy (>1-year old) to adulthood.
2 Our new feeding scale can be used by those who have never needed tube feeding, in addition to those who have, as long as 5% or more of their nutritional intake is through the oral route.
3 Our new feeding scale is validated for use in CHARGE syndrome, a genetic condition with a wide spectrum of clinical features. It has the potential to be validated in patient populations with many more genetic syndromes.

Feeding difficulties are reported to be one of the most complex and life-long problems in individuals with genetic syndromes.[1] Feeding problems are associated with severe morbidity and mortality in these populations.[1–7] The complexity of feeding problems is due to interactions among a multitude of dysfunctions such as cranial nerve anomalies, brain stem dysfunction, craniofacial abnormalities, uncoordinated swallowing, oral hypotonia and abnormal tongue movement.[1] In CHARGE syndrome (C = coloboma, H = heart defects, A = atresia choanae, R = retardation of growth, G = genitourinary anomalies, E = ear abnormalities), feeding problems often appear early in life, with over 90% of individuals needing some form of supplemental tube feeding.[8–10] Feeding difficulties, however, may manifest at any time in the individual's life, including adolescence and adulthood.[4,11–16] As CHARGE syndrome was only first described in 1979,[17,18] with the causative gene mutation not discovered

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Conflict of interest: None declared.

Accepted for publication 23 February 2021.

until 2004,[19] we are continually learning and discovering the complexity and uniqueness of the extensive feeding difficulties these individuals face.

Unique feeding difficulties that have been identified in CHARGE syndrome include severe gastroesophageal reflux and associated motility issues (requiring multiple therapies), mouth over-stuffing, pocketing of food in cheeks, high aspiration risk, and prolonged G-J tube feeding.[5,20–23] The Blake criteria[24] identified cranial nerve anomalies as a major component of CHARGE syndrome which has resulted in the suggestion of adding feeding and swallowing dysfunction to the clinical diagnostic criteria of CHARGE syndrome, indicating its high prevalence and importance.[23,25]

Existing paediatric Feeding Assessment Scales (FASs) developed for typically developing children may not accurately capture the feeding difficulties faced by those with genetic syndromes.[1] There is also no currently existing scale that can be used in both children who are tube fed, as well as those who are not. Lastly, existing scales are often limited to use in children and are not validated for continued use into adulthood, despite individuals with genetic syndromes often experiencing feeding issues well beyond their childhood. Therefore, the objective of the current study was to develop a new feeding scale for use in genetic syndromes across the life-span, and to assess its reliability and validity as a parent/care giver-report tool in individuals with CHARGE syndrome older than 1 year of age.

Methods

Participants

Ethics approval was received from the Research Ethics Board. Parents/care givers of individuals with CHARGE syndrome whose child was 1 year of age or older were invited to participate. Individuals had to receive some amount of their daily nutrition by oral feeding. Those with exclusive tube feeding were excluded.

Measures

Feeding Assessment Scale

The new FAS (Fig. 1) was developed by using the previously validated and reliable Pediatric Assessment Scale for Severe Feeding Problems (PASSFP) as a guide.[26] The PASSFP was designed to assess progress in the development of oral eating skills for children with neurological conditions who need prolonged tube feeding, and permission was obtained to adapt its format. An extensive review of the existing literature on the topic of feeding/gastrointestinal issues in genetic syndromes was used to adapt existing questions and to add new questions. Each question was individually reviewed and revised by four international experts in genetic conditions and feeding difficulties. Subsequently, 10 parents/care givers of children, adolescents and adults with CHARGE syndrome piloted the scale and provided feedback for each question. Based on this feedback, certain words were changed and revisions were added to the scale to improve question clarity and readability (e.g. changing the word 'problem' to 'difficulties', adding in the phrase 'on purpose or not'). After piloting the new scale, an additional question was added to the scale asking whether or not the individual had difficulty with very thin liquids (e.g. water) as parents/care givers felt this was a significant swallowing problem, and liquid intake represents the highest risk of aspiration in individuals with genetic syndromes.[27]

The new FAS is a 31-item parent-report measure. The first section (22 items) consists of questions related to how often certain feeding difficulties are happening in the individual's daily life (e.g. he/she/they will refuse food when eating orally). It is scored on a 5-point Likert scale: Never = 0, A little = 1, Sometimes = 2, A lot = 3, Always = 4. The second section (nine items) consists of questions related to the type of food the individual has difficulty with (e.g. cold foods, pureed foods). It is scored as No (0 points) or Yes (1 or 2 points). Difficulties with pureed foods (e.g. applesauce), mashed lumpy food (e.g. mashed vegetables) and soft chewable foods (e.g. bread) were scored 2 points for a 'Yes' answer, compared to the remainder of the second section that was scored with 1 point for a 'Yes'. The increased points assigned to these specific questions were based on existing literature that problems with these types of foods indicated more severe feeding difficulties than the other food options (e.g. hard vegetables).[2,3] The scale takes approximately 5 min to complete, and can easily be scored by the user immediately upon completion. Each of the 31 items is added up to a total score. Scores range from 0 to 100, with higher scores indicating more severe feeding difficulties.

The PASSFP is an existing valid and reliable scale designed to assess progress in the development of oral eating skills for children with neurological conditions who need prolonged tube feeding.[26] It is a 15-item parent report measure that covers nutritional topics, oral sensory issues, motor deficits, behavioural issues and quality of life issues. Scores range from 0 to 66, with lower scores representing more severe feeding difficulties.

Demographics

Parents/care givers were asked to report basic demographics (e.g. location, age) and the CHARGE syndrome characteristics of the individual with CHARGE syndrome (e.g. coloboma, choanal atresia). They were also asked a series of questions about the individual's feeding history (e.g. frequency of feeding therapy sessions per month, past/present tube feeding).

Feeding history questions

Based on the existing literature, six of the 'Yes/No' feeding history questions were used as markers of the severity of current feeding difficulties: (i) current tube feeding; (ii) currently in feeding therapy or needed to start soon; (iii) current weak sucking/chewing; (iv) current aspiration risk; (v) diet was currently limited to certain foods; and (vi) current safety concerns for feeding. For each individual, the number of 'yes' answers to each of these six feeding history questions was summed. This sum represented an approximation of how severe the individual's current feeding and swallowing difficulties were. Separate from the scale, parents/care givers also rated how they would best describe the individual's current feeding difficulties on a 6-point Likert scale (0 = Very mild, 1 = Mild, 2 = Mild to moderate, 3 = Moderate, 4 = Severe, 5 = Very severe). This method of severity rating was previously used in the PASSFP.[26]

Procedure

Given the low incidence of CHARGE syndrome served by any one medical centre, and due to the highly organised official Facebook pages created by official CHARGE syndrome foundations, participants were recruited from Facebook pages of the official International CHARGE Syndrome foundations. A secure

electronic link to the three questionnaires (see Materials) was distributed via Opinio software (ObjectPlant, Inc., Oslo, Norway). To examine test–retest reliability, parents were invited via email to complete the questionnaires a second time 4–8 weeks after their first submission.

Data analysis

All statistical analyses were conducted in SPSS Version 20.0.0 (IBM SPSS Statistics, 1 New Orchard Road, Armonk, New York, USA). All analyses were two-sided and a *P* value of 0.05 or less was deemed significant. Test–retest reliability was analysed using a paired samples

Feeding Assessment Scale (FAS)

Date: _____

Name of Individual: _____

Age:_____ Gender (Circle one): Male Female Not Disclosed

Completed By (Circle one): Mother Father Feeding Therapist Nurse/Physician Other:_____

What percentage of your child/adult's daily fluid/nutrition intake is by G/J tube feeding? (Circle one percentage):

0%	25%	50%	75%	95%

Circle one number on the scale:		Never	A Little	Sometimes	A lot	Always
1	He/she will refuse food when eating orally.	0	1	2	3	4
2	He/she takes longer than 45 minutes to eat orally.	0	1	2	3	4
3	He/she takes less than 15 minutes to eat orally.	0	1	2	3	4
4	He/she needs **close supervision** when eating orally.	0	1	2	3	4
5	He/she needs **someone in the room** when eating orally.	0	1	2	3	4
6	He/she has problems cutting food when eating orally.	0	1	2	3	4
7	He/she has problems feeding him/herself when eating orally.	0	1	2	3	4
8	He/she chokes or coughs when eating orally.	0	1	2	3	4
9	He/she has **trouble** chewing food.	0	1	2	3	4
10	He/she has **trouble** swallowing food.	0	1	2	3	4
11	He/she has to be **told or reminded** to chew.	0	1	2	3	4
12	He/she has to be **told or reminded** to swallow.	0	1	2	3	4
13	He/she does not like to mix food textures when eating (e.g. mixing puree and solid food).	0	1	2	3	4
14	He/she accidentally loses food out of his/her mouth during eating.	0	1	2	3	4
15	He/she will over-stuff his/her mouth with food during eating.	0	1	2	3	4

Fig 1 The new feeding assessment scale (FAS).

Circle one number on the scale:		Never	A Little	Sometimes	A lot	Always
16	He/she has difficulty moving food around with his/her tongue during eating.	0	1	2	3	4
17	He/she has a hard time feeling food or anything touching the inside of his/her mouth.	0	1	2	3	4
18	He/she dislikes oral eating.	0	1	2	3	4
19	He/she lets food sit in his/her **cheeks or palate** during eating (on purpose or not).	0	1	2	3	4
20	He/she will have food hidden in his/her **cheeks or palate** after the meal has ended (on purpose or not).	0	1	2	3	4
21	The **Parent/Caregiver** gets worried about their child/adult's ability to eat orally.	0	1	2	3	4
22	The **Parent/Caregiver** has difficulties feeding their child/adult. (e.g. preparing food the right way, getting enough information about helping them eat/drink)	0	1	2	3	4
Does the child/adult have problems with:		**No**		**Yes**		
23	Cold foods	0		1		
24	Room temperature foods	0		1		
25	Warm foods	0		1		
26	Very thin liquids (e.g. water)	0		1		
27	Pureed foods (e.g. applesauce)	0		2		
28	Mashed lumpy food (e.g. mashed potatoes or mashed vegetables)	0		2		
29	Soft chewable foods (e.g. bread, crackers)	0		2		
30	Tough chewable foods (e.g. meat)	0		1		
31	Hard vegetables and fruit (e.g. raw apples)	0		1		
Total Score (sum of all items)		**/100 total points**				

Fig 1 Continued

t-test. A univariate analysis of variance was used to assess construct validity and face validity. Internal validity was assessed by Pearson's correlation between the first and second components of the scale as well as Cronbach's alpha. Convergent validity was analysed using Pearson's correlation between the new feeding scale and the existing validated PASSFP scale. Feeding scale scores were compared between individuals with and without certain feeding difficulties and CHARGE syndrome characteristics using an independent samples *t*-test. These

feeding difficulties included 'Yes/No' questions of the following: current tube feeding (any type), gastrostomy tube feeding (past or present), jejunostomy tube feeding (past or present), nasogastric tube feeding (past or present), aspiration, weak sucking/chewing, safety concerns for feeding, diet limited to certain foods (e.g. puree), currently in feeding therapy, not in feeding therapy currently but needs to start soon, tracheostomy in the past, gastroesophageal reflux (current) and surgeries to improve feeding/swallowing (e.g. Nissen fundoplication) (see Table 5). One of the authors is a consulting scientist for the Research Services Department at the local hospital and has expertise in biostatistics. She reviewed all the statistical methods employed and results in this manuscript.

Results

Participants

One hundred parents/care givers (92 mothers) participated. Individuals with CHARGE syndrome were evenly distributed by gender (48 females) and had ages that ranged from 1 to 33 years old, with a mean age of 11 ± 8 years. Individuals were from a varied geographical distribution including the USA ($n = 61$), UK ($n = 15$), Australia ($n = 9$), Canada ($n = 6$), New Zealand ($n = 2$), Belgium ($n = 1$) and Estonia ($n = 1$).

All 100 individuals had a clinical diagnosis of CHARGE syndrome from a clinician using the Blake[24] and/or Verloes [28] diagnostic criteria. Sixty-two of the 100 individuals had their clinical diagnosis additionally confirmed with genetic testing. The mean

Table 1 Frequency of major and minor phenotypic CHARGE syndrome features in the study population ($n = 100$)

	%	General CHARGE syndrome population frequency,[9] %
Major CHARGE syndrome features		
External ear abnormalities	84	95–100
Middle ear abnormalities	82	—
Inner ear abnormalities	90	90
Abnormal semicircular canals	69	—
Coloboma (retina/iris)	73	75–90
Choanal stenosis/atresia	56	65
Cranial nerve dysfunction	96	—
CN I (difficulty smelling)	39	—
CN VII (facial palsy)	52	50–90
CN VIII (difficulty hearing)	88	—
CN IX, X, XI (difficulty swallowing)	53	—
Minor CHARGE syndrome features		
Distinct CHARGE facial features	77	—
Developmental delay	91	>70
Heart defect	73	50–85
Cleft lip or palate	24	15–20
Cleft lip	19	—
Cleft palate	23	—
Genital hypoplasia	41	50–70
Growth deficiency	35	—
Tracheoesophageal fistula	25	—
Urinary tract dysfunction	16	—

Table 2 Feeding history characteristics of 100 individuals with CHARGE syndrome

	%
Never had tube feeding†	20
Past tube feeding only†	47
Current tube feeding†	33
Mean age that tube feeding† began (for past or current users), months	4 ± 12
Mean age that tube feeding† stopped (for past users), years	5 ± 5
Types of previous/current tube feeding‡	
Nasogastric	35
Gastrostomy	63
Jejunostomy	13
Oral-gastric tube	4
Had surgery to improve feeding or swallowing§	42
Had a tracheostomy	18
Feeding Therapy	
Never had feeding therapy	14
Past feeding therapy but not current	64
Currently in feeding therapy	22
Feeding therapists seen¶	
Speech language pathologist	76
Occupational therapist	56
Other (e.g. Dietician)	11
Psychologist	4
Weak sucking or chewing skills diagnosed by clinician	33
Gastroesophageal reflux (GER)	
Never a problem	16
A problem in the past only	40
Currently taking medication for GER	44
Aspiration††	
Never a problem	27
A problem in the past only	51
Currently a problem	22
Current diet has limitations on texture or type of food‡‡	45
Current safety concerns§§ with respect to feeding	30

†Tube feeding = Includes any of nasogastric, gastrostomy, jejunostomy, oral-gastric tube feeding.

‡Individuals were able to select one or more types of tube feeding that they had previously/currently.

§For example, Nissen fundoplication, tracheoesophageal fistula repair.

¶Individuals were able to select one or more types of feeding therapists seen.

††Aspiration diagnosed by a clinician (e.g. by barium swallow, recurrent pneumonias etc.) was reported by parents.

‡‡For example, diet is limited to puree texture only, limited to only soft foods, etc.

§§For example, risk of choking.

Table 3 Number one feeding/swallowing difficulty reported by parents of 100 individuals with CHARGE syndrome

	%
Swallowing	25
Chewing	19
Mouth over-stuffing while eating	16
Food texture aversion	11
Drinking thin liquids	5
Gagging or choking	4
Does not like to eat or try new food	4
Aspiration	3
Motor control†	2
Eating independently	2
Anatomical obstruction‡	2
No response provided	7

†For example, cannot control the movement of food inside mouth, loses food out of mouth easily.
‡For example, oesophageal narrowing.

age of diagnosis (clinical or genetic) was 1 year of age (standard deviation 2, range 0–11 years). The frequencies of major and minor CHARGE syndrome characteristics of this study population were all within the known frequencies in the general CHARGE syndrome population (Table 1).

Feeding history characteristics

Parent/care giver-reported feeding difficulties of the individuals with CHARGE syndrome in our study are displayed in Table 2. Problems with swallowing, chewing, and mouth over-stuffing while eating were the most common feeding difficulties reported (Table 3).

Test–Retest reliability

Thirty parents/care givers (30%) completed the scale a second time approximately 4–8 weeks (M = 43 ± 28 days) after the first completion to measure test–retest reliability. The mean score of the first administration (32 ± 17) and the second administration (31 ± 17) were not significantly different (t = 1.4, df = 29, P = 0.2, CI −0.8, 3.8). The test–retest correlation was high at 0.94 ($P < 0.001$).

Construct validity

There was a significant effect for number of feeding difficulty risk factors on the new FAS scores, with increased feeding risk factors having higher scale scores (F = 10.9, df = 6, 98, $P < 0.001$). Mean scores for the number of feeding difficulty risk factors were as follows: zero (M = 27 ± 13), one (M = 36 ± 18), two (M = 43 ± 13), three (M = 46 ± 8), four (M = 58 ± 19), five (M = 50 ± 14) and six (M = 61 ± 15) risk factors. A Tukey post-hoc test revealed that individuals who had zero feeding risk factors had significantly lower FAS scores than individuals who had two, three, four, five or six risk factors ($P < 0.03$). There was no significant effect of number of feeding difficulty risk factors on the age of the individual with CHARGE syndrome (F = 1.3, df = 6, 98, P = 0.250). Based on these results, three groups were created using the FAS scores: mild (0–25 points), moderate (26–50 points) and severe (51–100) feeding difficulties.

Internal validity

The first (5-point Likert scale) and second section (Yes/No) total scores significantly correlated with each other (r = 0.601, $P < 0.001$). Cronbach's alpha was 0.91, indicating a high level of internal consistency.

Face validity

There was a significant effect for the six groups of parents/care givers' descriptions of their child's current feeding difficulties (very mild to very severe) on the FAS total scores (F = 37.6, df = 5, $P < 0.001$) and PASSFP total scores (F = 31.0, df = 5, $P < 0.001$) (Table 4). For the FAS, all parent-reported categories differed significantly in mean scores except for 'mild-to-moderate' compared to 'moderate'. For the PASSFP scale, all categories differed significantly except for 'very mild' compared to 'mild', as well as 'mild to moderate' compared to 'moderate'.

Convergent validity

Convergent validity was analysed between the FAS and the PASSFP. There was a significant negative correlation between the

Table 4 New Feeding Assessment Scale (FAS) and Pediatric Assessment Scale for Severe Feeding Problems (PASSFP) scores for individuals with CHARGE syndrome based on parent-reported severity of current feeding difficulties

Parent's subjective description of individual's current feeding difficulties	*n*	Mean FAS score (SD)	Mean PASSFP score (SD)
Very mild†,‡	14	19 (±11)	55 (±5)
Mild§,¶	22	27 (±9)	52 (±6)
Mild to moderate††,‡‡	14	39 (±11)	46 (±7)
Moderate§§,¶¶	14	42 (±7)	45 (±3)
Severe†††,‡‡‡	18	50 (±8)	40 (±8)
Very severe	18	63 (±17)	28 (±10)

Equal variances not assumed. New FAS and PASSFP total scores:
†Compared with mild, t = −2.3, df = 23.4, P = 0.029*.
‡Compared with mild, t = 1.5, df = 31.6, P = 0.133.
§Compared with mild to mod, t = −3.7, df = 22.9, P = 0.001*.
¶Compared with mild to mod, t = 2.7, 26.3, P = 0.013*.
††Compared with mod, t = −0.9, df = 22.3, P = 0.361.
‡‡Compared with mod, t = 0.175, df = 18.0, P = 0.863.
§§Compared with severe, t = −3.0, df = 29.2, P = 0.005*.
¶¶Compared with severe, t = 2.6, df = 21.8, P = 0.018*.
†††Compared with very severe, t = −3.0, df = 23.8, P = 0.006*.
‡‡‡Compared with very severe, t = 3.9, df = 30.3, $P < 0.001$*.
SD, standard deviation.

two scales ($r = -0.785$, $n = 97$, $P < 0.001$). Higher scores on the FAS and lower scores on the PASSFP both indicate more severe feeding difficulties.

Parent/care giver-reported feeding difficulties

Table 5 displayed the comparisons of FAS total scores between individuals with a specific feeding difficulty and those without that feeding difficulty. FAS total scores were significantly worse (higher) for individuals who replied 'Yes', compared to individuals who replied 'No' for the following feeding difficulties: current tube feeding, gastrostomy/jejunostomy/nasogastric tube feeding (past or present), aspiration, weak sucking/chewing, safety concerns for feeding, diet limited to certain foods, currently in feeding therapy, needing to start feeding therapy soon, and history of tracheostomy. The only two feeding history questions in which FAS scores of those who responded 'Yes' did not significantly differ from individuals who responded 'No' were: (i) current gastroesophageal reflux; and (ii) if the individual had undergone previous surgeries (e.g. Nissen fundoplication) to improve feeding/swallowing (Table 5).

CHARGE syndrome characteristics

Individuals with cranial nerve IX, X, XI dysfunction (swallowing difficulties) had significantly higher FAS total scores (44 ± 19) than those without (29 ± 14) ($P = 0.002$) (Table 6). Individuals with a cleft palate also had significantly higher total scores (47 ± 22) than those without a cleft palate (38 ± 16) ($P = 0.03$). For all other CHARGE syndrome features, FAS scores did not significantly differ between individuals with and without a certain feature.

Discussion

Our study revealed that the newly developed FAS is a reliable and valid parent/care giver-report tool in individuals with CHARGE syndrome older than 1 year old. The test–retest reliability was high. Validity was demonstrated through high internal validity. The face validity of our scale was also high as scale scores differed significantly depending on how severe the parents/care givers reported their individual's feeding issues: very mild, mild, moderate, severe, and very severe. The FAS was better at detecting differences between milder feeding difficulties than the PASSFP scale, whose scores did not differ between very mild and

Table 5 Presence or absence of specific feeding difficulties and their relationship to the new Feeding Assessment Scale (FAS) mean total score in 100 individuals with CHARGE syndrome

Feeding difficulty	%	FAS mean scores (SD)	*P* value†	95% confidence interval
Current tube feeding (any type)	Yes (33)	53 (±16)	<0.001*	−24.3, −10.8
	No (67)	34 (±16)		
Gastrostomy tube feeding (past or present)	Yes (61)	43 (±19)	0.02*	−15.3, −1.2
	No (39)	35 (±16)		
Jejunostomy tube feeding (past or present)	Yes (13)	51 (±18)	0.04*	−23.9, −0.8
	No (87)	39 (±18)		
Nasogastric tube feeding (past or present)	Yes (34)	45 (±17)	0.04*	−15.1, −0.3
	No (66)	38 (±18)		
Aspiration	Yes (22)	51 (±16)	0.001*	−22.1, −5.9
	No (78)	37 (±18)		
Weak sucking/chewing	Yes (33)	53 (±16)	<0.001*	−27.8, −15.0
	No (67)	32 (±15)		
Safety concerns for feeding	Yes (30)	51 (±17)	<0.001*	−23.3, −8.5
	No (70)	35 (±17)		
Diet limited to certain foods (e.g. puree)	Yes (45)	48 (±18)	<0.001*	−21.0, −7.5
	No (55)	34 (±16)		
Currently in feeding therapy	Yes (22)	53 (±19)	0.001*	−25.5, −7.7
	No (78)	36 (±15)		
Not in feeding therapy currently but needs to start soon	Yes (12)	50 (±13)	0.002*	−25.0, −7.0
	No (88)	34 (±15)		
Tracheostomy in the past	Yes (18)	51 (±16)	0.005*	−21.3, −4.2
	No (82)	38 (±18)		
Gastroesophageal reflux (current)	Yes (37)	41 (±18)	0.6	−9.4, 5.7
	No (63)	39 (±19)		
Surgeries to improve feeding/swallowing (e.g. Nissen fundoplication)	Yes (42)	42 (±20)	0.4	−10.5, 4.6
	No (58)	39 (±17)		

†Yes and No groups mean scores were compared with an independent-samples *t*-test (two-tailed). *P* value of <0.05 was significant.
SD, standard deviation.

Table 6 CHARGE syndrome characteristics and their relationship to the new Feeding Assessment Scale (FAS) mean total score in 100 individuals with CHARGE syndrome

	%	FAS mean scores (SD)	*P* value†	95% confidence interval
Major CHARGE syndrome features				
Abnormal semicircular canals	Yes (69)	39 (±16)	0.4	−26.1, 10.5
	No (31)	32 (±25)		
Coloboma (iris/retina)	Yes (73)	41 (±18)	0.8	−9.9, 7.4
	No (27)	39 (±20)		
Choanal atresia/stenosis	Yes (56)	39 (±19)	0.4	−4.0, 10.5
	No (44)	42 (±18)		
Cranial nerve I dysfunction‡ (absent sense of smell)	Yes (39)	38 (±18)	0.3	−7.3, 27.3
	No (61)	48 (±29)		
Cranial nerve VII dysfunction‡ (facial palsy)	Yes (52)	41 (±17)	0.2	−13.5, 2.6
	No (48)	36 (±19)		
Cranial nerve VIII dysfunction‡ (hearing impairment)	Yes (88)	40 (±19)	0.9	−17.1, 20.6
	No (12)	41 (±8)		
Cranial nerve IX, X, XI, XII dysfunction‡ (swallowing difficulties)	Yes (53)	44 (±19)	0.002*	−25.3, −5.9
	No (47)	29 (±14)		
Minor CHARGE syndrome features				
Developmental delay	Yes (91)	40 (±18)	0.2	−27.9, 5.1
	No (9)	29 (±11)		
Heart defect	Yes (73)	40 (±19)	0.9	−7.2, 7.6
	No (27)	40 (±15)		
Cleft lip	Yes (19)	45 (±22)	0.2	−15.5, 3.0
	No (81)	39 (±17)		
Cleft palate	Yes (23)	47 (±22)	0.03*	−18.0, −1.2
	No (77)	38 (±16)		
Genital hypoplasia	Yes (41)	41 (±17)	0.4	−11.5, 4.8
	No (59)	38 (±20)		
Growth deficiency	Yes (35)	39 (±19)	1.0	−9.4, 9.3
	No (65)	39 (±17)		
Tracheoesophageal fistula	Yes (25)	43 (±19)	0.3	−13.8, 3.7
	No (75)	38 (±19)		
Urinary tract dysfunction	Yes (16)	41 (±17)	0.7	−11.7, 8.4
	No (84)	39 (±19)		

†Yes and No groups mean scores were compared with an independent-samples *t*-test (two-tailed). *P* value of <0.05 was significant.
‡Cranial nerve dysfunction was by parent report of clinician's diagnosing-specific cranial nerve dysfunction through neurological examination.
SD, standard deviation.

mild report groups. The FAS was also found to have significant convergent validity with the reliable and valid PASSFP scale, indicating that the FAS was indeed assessing feeding difficulties in the study population. However, the significant correlation did not approach the value of 1, indicating that there were differences in the assessment tools, which is what was expected as we had adapted the PASSFP questions to include questions regarding adolescents and adults with genetic syndromes, as well as questions regarding feeding difficulties experienced by those who had never used tube feeding or who were not currently tube feeding. To demonstrate the utility of the FAS, two case histories are presented of same-aged individuals with similar CHARGE syndrome characteristics, differing in their severity of feeding difficulties (Table 7).

To date, feeding and swallowing difficulties in CHARGE syndrome have primarily focused on gastroesophageal reflux disease (GERD). This diagnosis is common and individuals often undergo Nissen fundoplication.[2,3] These surgical procedures have had a low success rate in this population and individuals often need lifelong medical management.[29] However, other feeding difficulties have often been overlooked in favour of GERD. Interestingly, our study showed that individuals with or without GERD did not differ significantly in their severity of feeding difficulties based on the new feeding scale scores. GERD was the only feeding/swallowing difficulty that demonstrated this, as individuals who said yes to any other feeding/swallowing difficulty (e.g. aspiration, weak sucking/chewing, etc.) had significantly more severe feeding difficulties (higher feeding scale scores). Efforts should focus on assessing other areas of feeding and swallowing (e.g. if their suck/chew is weak or if there are signs of aspiration), as these can more accurately convey the severity of swallowing and feeding issues among individuals with CHARGE.

Our study also identified that two CHARGE syndrome characteristics were associated with significantly worse feeding

Table 7 Two case histories of same-aged individuals with similar CHARGE syndrome characteristics, differing in their severity of feeding difficulties

Mild feeding difficulties
An 11-year-old female received a clinical diagnosis of CHARGE syndrome, given at 2 weeks of age. Major diagnostic criteria included unilateral choanal atresia, as well as external, middle, and inner ear abnormalities. Cranial nerve dysfunction included unilateral facial palsy (CN VII), bilateral hearing loss (CN VIII), and swallowing problems (CN IX, X, XI). Minor diagnostic criteria were a minor heart defect, growth deficiency, tracheoesophageal fistula, and developmental delay
Her parent-reported primary feeding difficulty was difficulty chewing meat. In the past, she had required gastrostomy tube feeding from age two months to four years. She had received two years of feeding therapy with a speech language pathologist. Gastroesophageal reflux and aspiration were both problems in the past, but not presently. Her current diet was not limited in any way due to safety concerns. She scored an 8 on the 100-point FAS, indicating mild feeding difficulties at that present time in her life
Severe feeding difficulties
An 11-year-old female received a genetic diagnosis of CHARGE syndrome at age 6 months. Major diagnostic criteria included bilateral coloboma of the retina, unilateral choanal stenosis, external, middle, and inner ear abnormalities, and abnormal semicircular canals. Cranial nerve dysfunction included unilateral facial palsy (CN VII), bilateral hearing loss (CN VIII), and swallowing problems (CN IX, X, XI). Minor diagnostic criteria were a minor heart defect, growth deficiency, and developmental delay
Her parent-reported primary feeding difficulty was swallowing difficulties. She had required tube feeding since birth. In the past, she had needed tube feeding through nasogastric, jejunostomy, and gastrostomy tubes. She was currently using a gastrostomy tube for 90% of her daily nutritional intake. She had undergone three Nissen fundoplication surgical procedures to treat severe gastroesophageal reflux and also had a tracheostomy. Aspiration remained an ongoing risk, and as such, her diet was limited. Her parent reported that she did not cope well with drinks and persistently coughed with food; therefore, she could only handle a maximum of two teaspoons of food per meal. Recurrent aspiration had resulted in bronchiectasis and pneumonia. She thoroughly enjoyed the social aspect of family meals, and loved to taste small amounts of pureed foods. She scored an 87 on the 100-point FAS, indicating severe feeding difficulties at that present time in her life

difficulties and higher scale scores. These two characteristics were the lower cranial nerve (IX, X, XI, XII) dysfunction (the key clinical problem being swallowing abnormalities), and cleft palates. A previous study on feeding difficulties in children with CHARGE syndrome also found cranial nerve dysfunction to be the primary clinical feature impacting feeding development.[9] It is essential that a comprehensive cranial nerve examination be performed, in order to identify any areas of weakness and to predict resulting swallowing/feeding difficulties.[10] This may include high-resolution magnetic resonance imaging (MRI) in certain cases. Cranial nerve IX, X, XI and XII all play essential roles in the sensory and motor innervation of the tongue, pharynx and larynx, contributing to the manipulation of food, formation of food bolus, and initiation of swallowing. Innervation may be dysfunctional along the entire GI tract, with reduced enteric system innervation and gut motility having been reported in zebrafish models of CHARGE syndrome.[30] A higher percentage of cleft palates have been previously reported in individuals with CHARGE syndrome who have the problematic feeding behaviours of pocketing food in cheeks and overstuffing food into their mouth, compared to the general CHARGE syndrome population.[21] The presence of a cleft palate as an infant can impair sucking behaviour due to an inability to close the nasal cavity. This disruption in learned sucking behaviour and delayed introduction of functional oral feeding may have a lasting impact on feeding issues throughout the individual's life, even after surgical cleft palate repair.[2,3] A previous review on feeding and swallowing dysfunction in genetic syndromes also found that craniofacial abnormalities lead to poor feeding and aspiration.[1]

The FAS has three primary applications. First, parents/care givers can use the FAS as a structured assessment of the current severity of feeding difficulties. Second, it can be used to track the individual's oral feeding progress before, during, and after feeding treatment (e.g. feeding therapy) or otolaryngological procedures, as well as across their entire life. Finally, by helping identify specific feeding problems, it can alert the patient's health-care team and feeding therapists to areas of concern.

Limitations and future research

The limitations of this current study involve the use of parental recall, with an inability to verify medical and feeding histories. Limitations also include that face validity was assessed with extra parent/care giver report questions, and not a validated tool/scale. However, the parents/care givers have expertise with CHARGE syndrome and have been advocates in bringing feeding issues to the professional's attention. Limitations to the FAS include its novelty and the requirement of further validation among clinicians and feeding therapists, as well as in individuals with other genetic syndromes. This new tool can also not be used by parents/care givers of individuals less than 1 year of age, as our study population excluded this group. Future research should continue to assess this tool in large populations, and in individuals with other genetic syndromes.

Conclusions

Our newly developed feeding scale is an easy-to-navigate tool that is valid and reliable in individuals with CHARGE syndrome. This scale can be used by parents/care givers of individuals 1 year of age or older who have begun to attempt oral feeding, and can be completed multiple times over the life-span to assess the improvement of feeding and swallowing difficulties. It can also alert the individual's care givers to new feeding problems that may develop, aiding them in voicing their concerns to the clinician or feeding therapist. Most importantly, this new tool can be easily completed and scored by any care giver familiar with the individual, although future research is needed for use beyond the primary care giver/parent. This feeding scale has the potential to be tested for validity and reliability in other genetic syndromes experiencing a high prevalence of otolaryngological and feeding problems. This feeding scale found that individuals with CHARGE

syndrome who had cleft palates and/or cranial nerve dysfunction had more significant severe feeding difficulties. This new tool can provide useful information for feeding therapists and other clinicians who are involved in the care of these individuals – from infancy (>1 year old) to adolescence, and beyond.

Acknowledgements

We would like to thank all of the wonderful families and individuals with CHARGE syndrome who made this work possible. This research was supported by the International CHARGE Syndrome Foundation and a summer studentship awarded by the Research in Medicine programme. This work was supported by Dalhousie University Research in Medicine Grant.

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