

Speech and language in *DYRK1A* syndrome



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ARTICLE

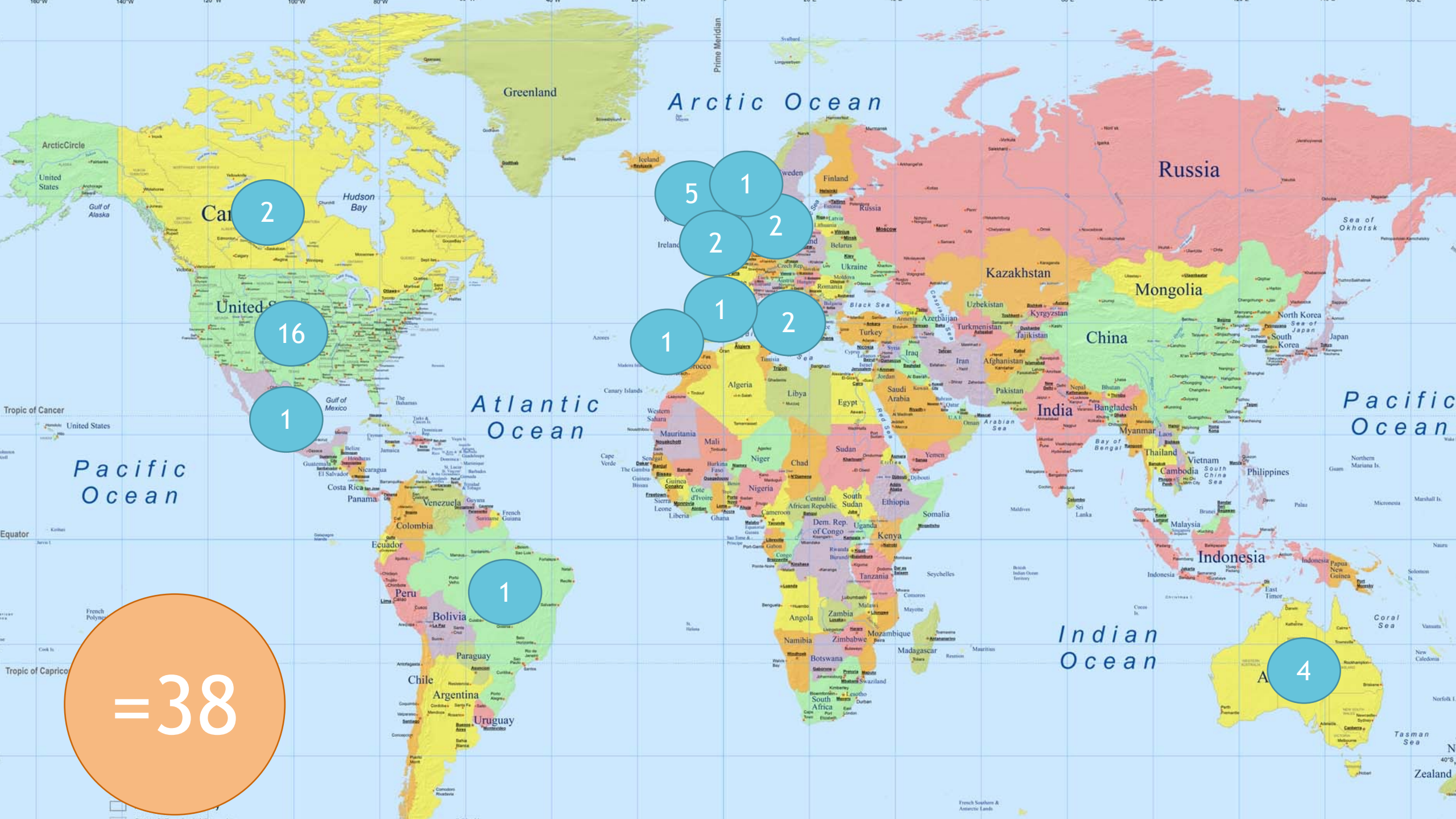


Social motivation a relative strength in DYRK1A syndrome on a background of significant speech and language impairments

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Speech and language impairments are commonly reported in *DYRK1A* syndrome. Yet, speech and language abilities have not been systematically examined in a prospective cohort study. Speech, language, social behaviour, feeding, and non-verbal communication skills were assessed using standardised tools. The broader health and medical phenotype was documented using caregiver questionnaires, interviews and confirmation with medical records. 38 individuals with *DYRK1A* syndrome (23 male, median age 8 years 3 months, range 1 year 7 months to 25 years) were recruited. Moderate to severe intellectual disability (ID), autism spectrum disorder (ASD), vision, motor and feeding impairments were common, alongside epilepsy in a third of cases. Speech and language was disordered in all participants. Many acquired some degree of verbal communication, yet few (8/38) developed sufficient oral language skills to rely solely on verbal communication. Speech was characterised by severe apraxia and dysarthria in verbal participants, resulting in markedly poor intelligibility. Those with limited verbal language (30/38) used a combination of sign and graphic augmentative and alternative communication (AAC) systems. Language skills were low across expressive, receptive, and written domains. Most had impaired social behaviours (25/29). Restricted and repetitive interests were most impaired, whilst social motivation was a relative strength. Few individuals with *DYRK1A* syndrome use verbal speech as their sole means of communication, and hence, all individuals need early access to tailored, graphic AAC systems to support their communication. For those who develop verbal speech, targeted therapy for apraxia and dysarthria should be considered to improve intelligibility and, consequently, communication autonomy.



2

16

1

1

5

1

2

2

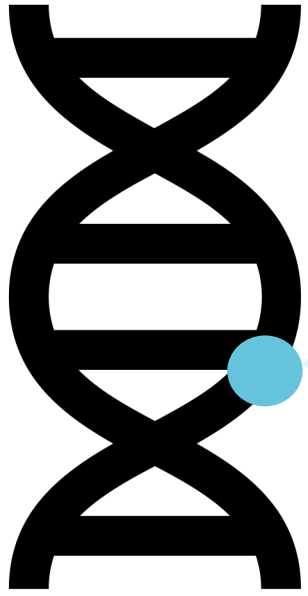
1

2

1

= 38

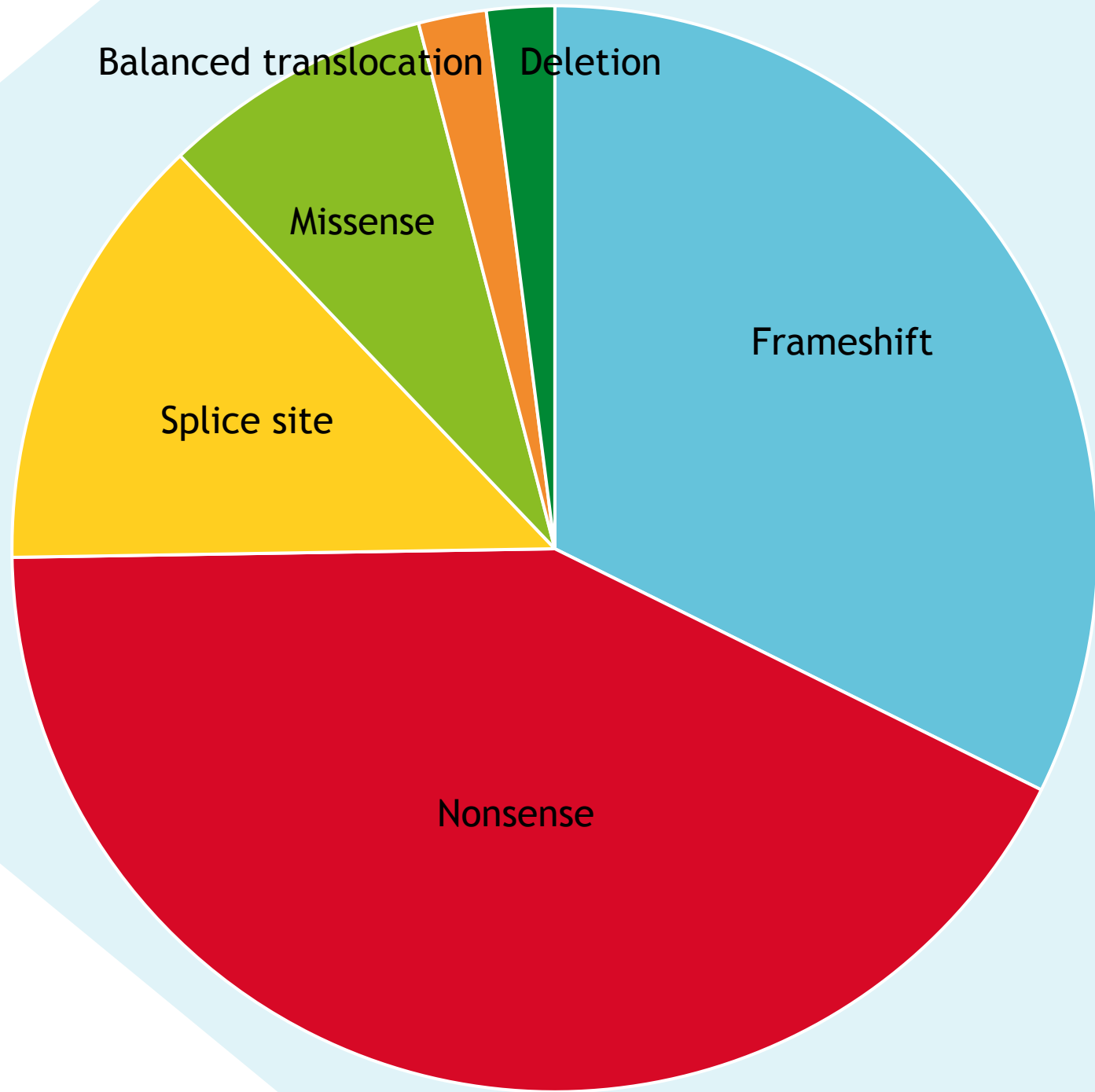
4



1 year



25 years



Speech

Learning & saying speech sounds

How many sounds do you have?

Do you have all the sounds of the language for their age?

Are the speech sounds clear?

Diagnosis:

- Articulation disorder
- Phonological disorder
- Phonological delay
- Stuttering
- Childhood Apraxia of Speech (CAS)
- Dysarthria

Language

Vocabulary: words you know & use

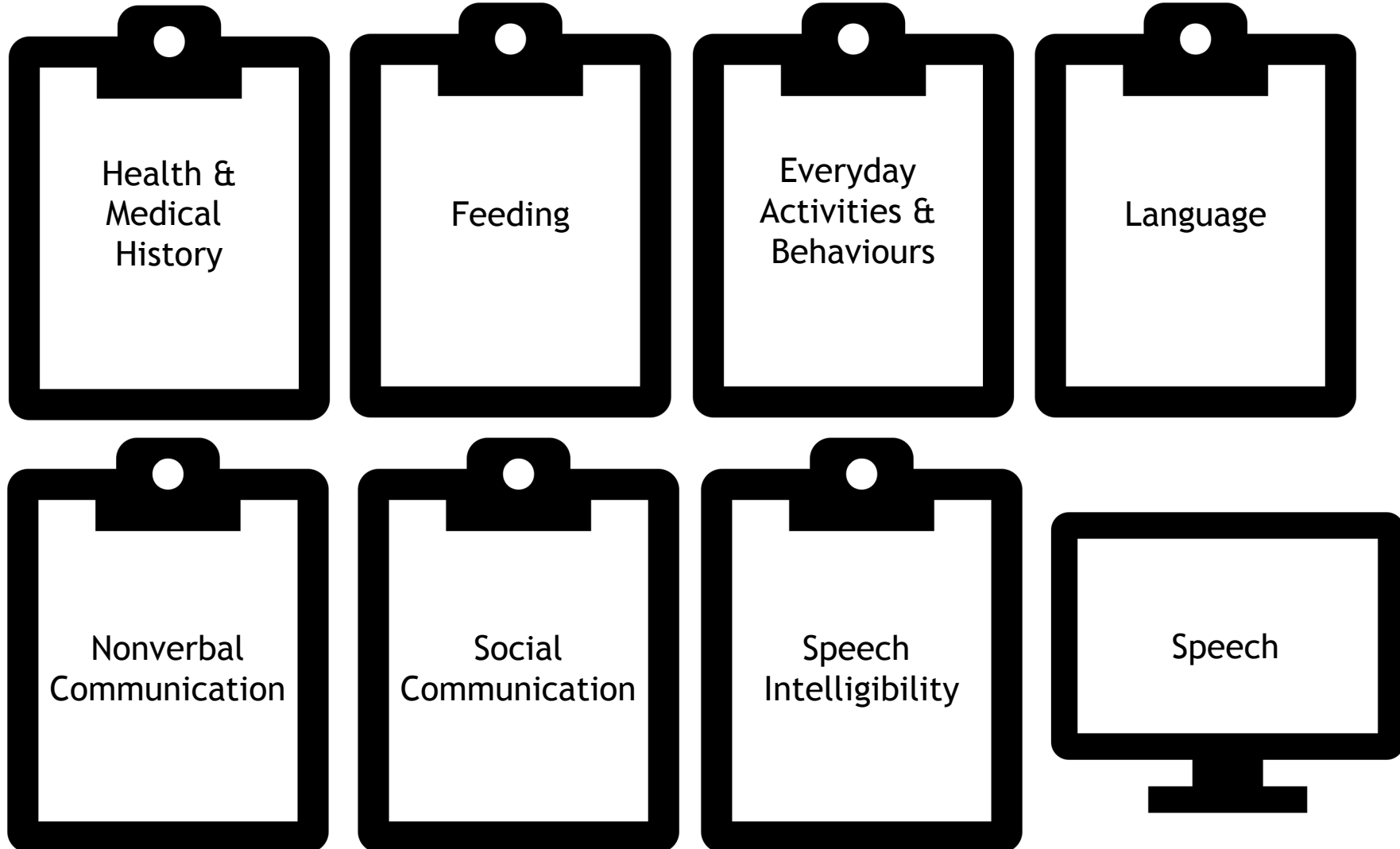
Grammar: how you make sentences

- Me wented to shop
- I went to the shops

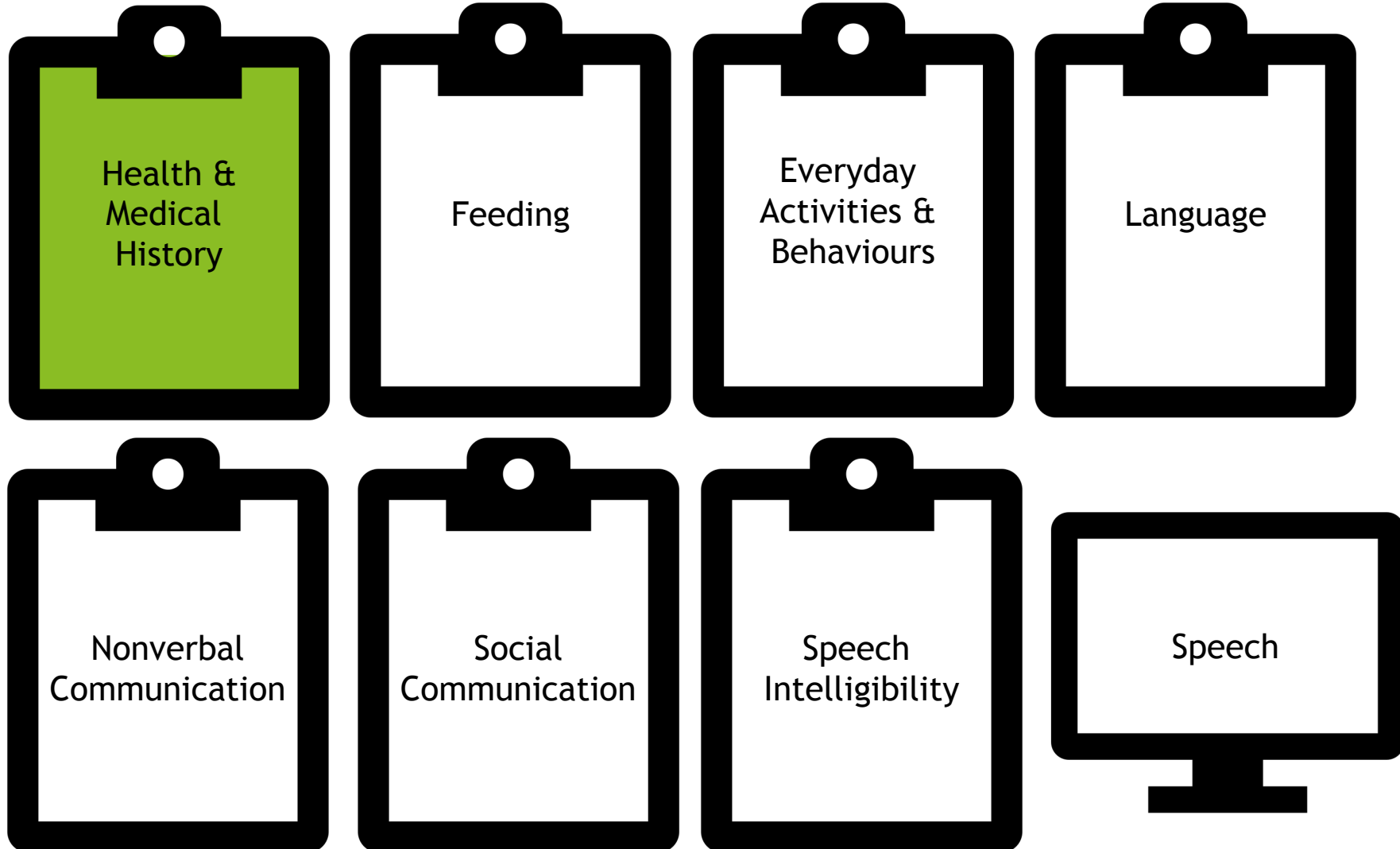
Appropriate for age?

Understanding (receptive) vs Use of language (expressive)

What did we assess?



What did we assess?

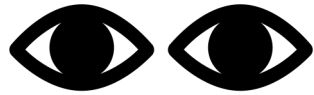


Health Conditions

20/38



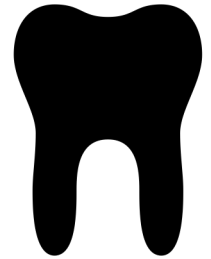
31/38



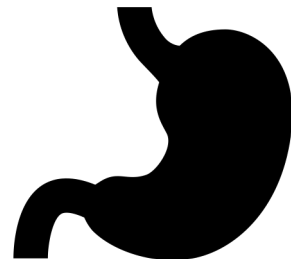
21/38



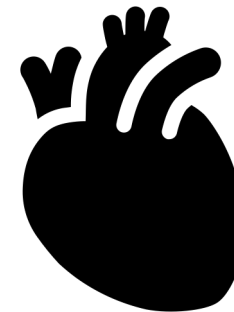
13/31



Microcephaly
Retro/Micrognathia

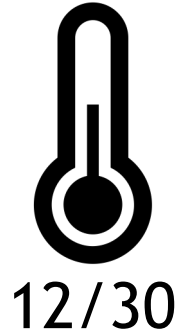
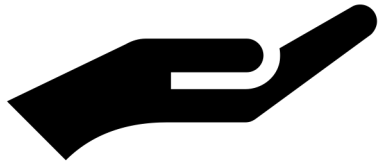


Constipation



9/38

OT: 33/38
PT: 33/38

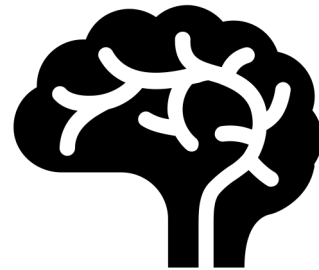
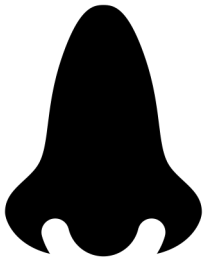


12/30



11/38

11/38

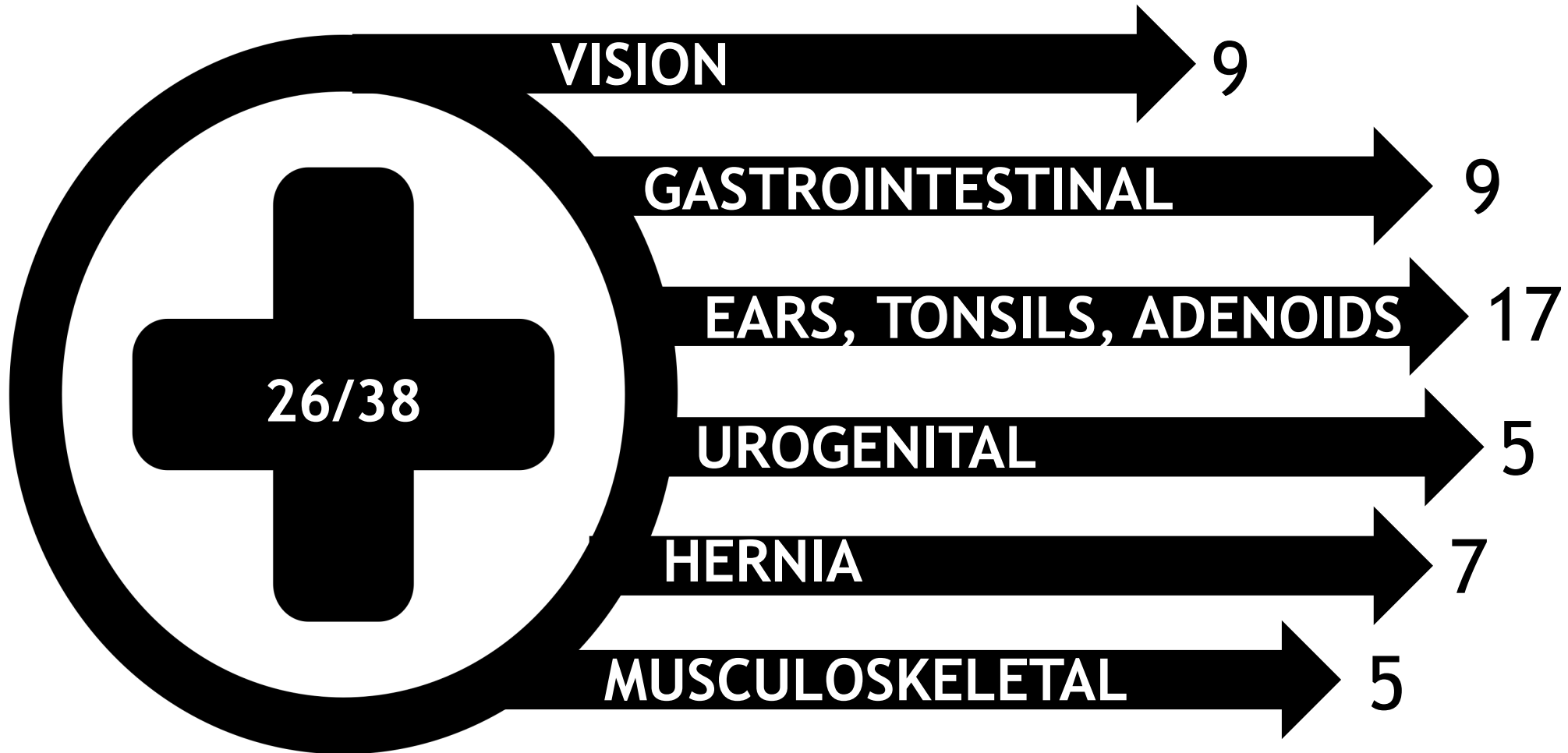


17/36



23/28

Surgeries

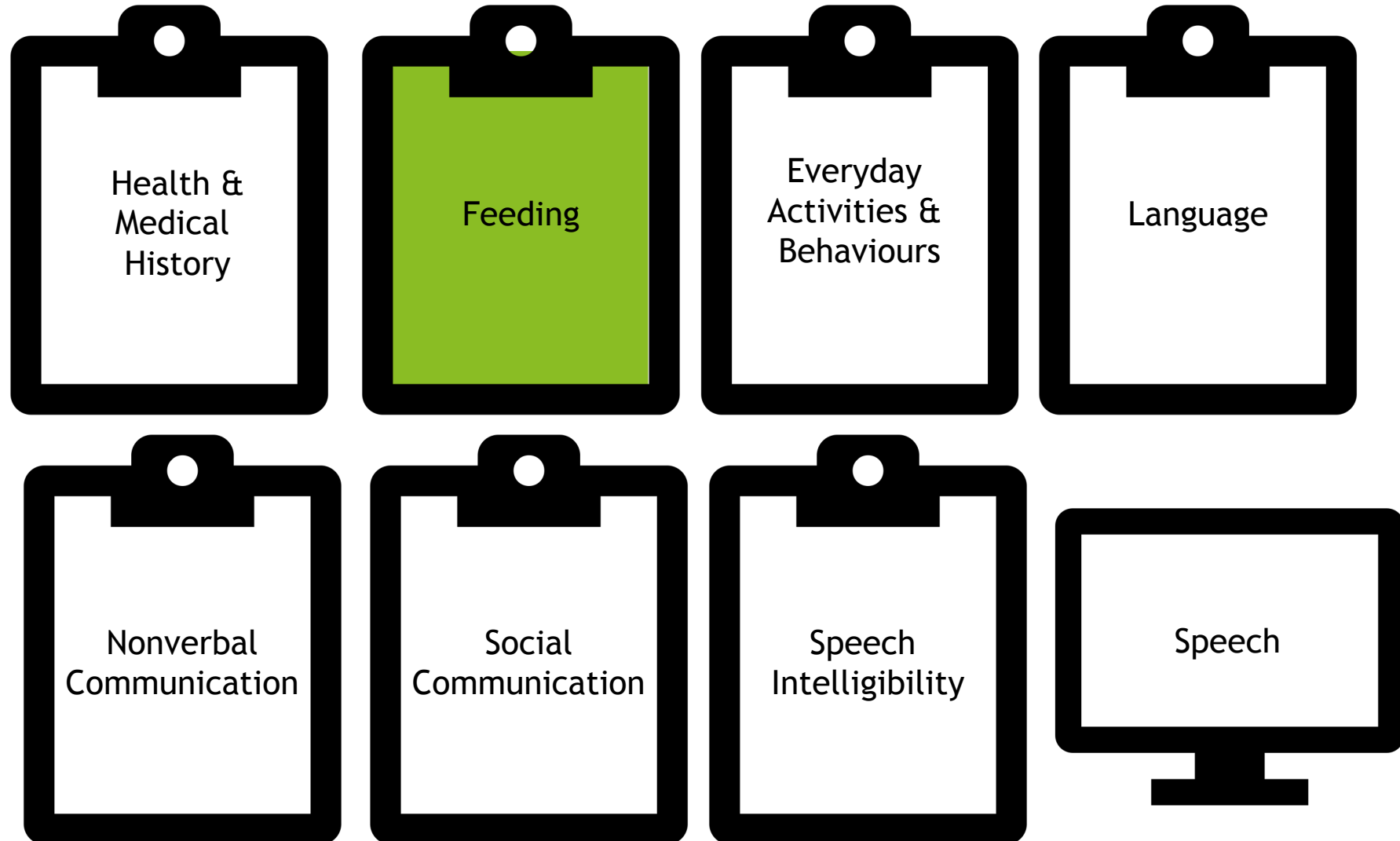


Neurodevelopmental Conditions



- Mild - Severe Intellectual Disability (28/28)
- Autism Spectrum Disorder (20/38)
- Attention Deficit Hyperactive Disorder (10/38)
- Other behavioural challenges

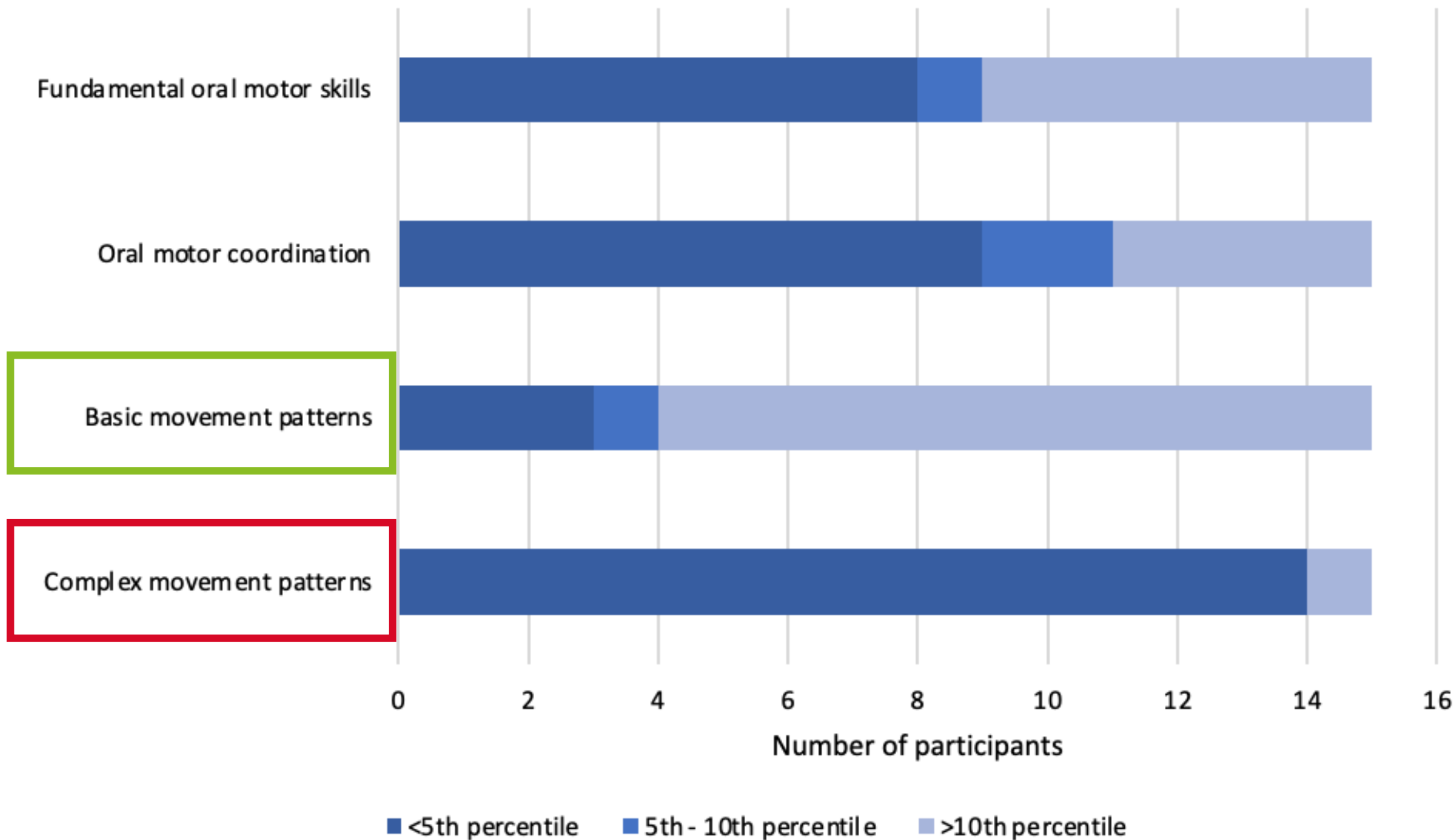
What did we assess?



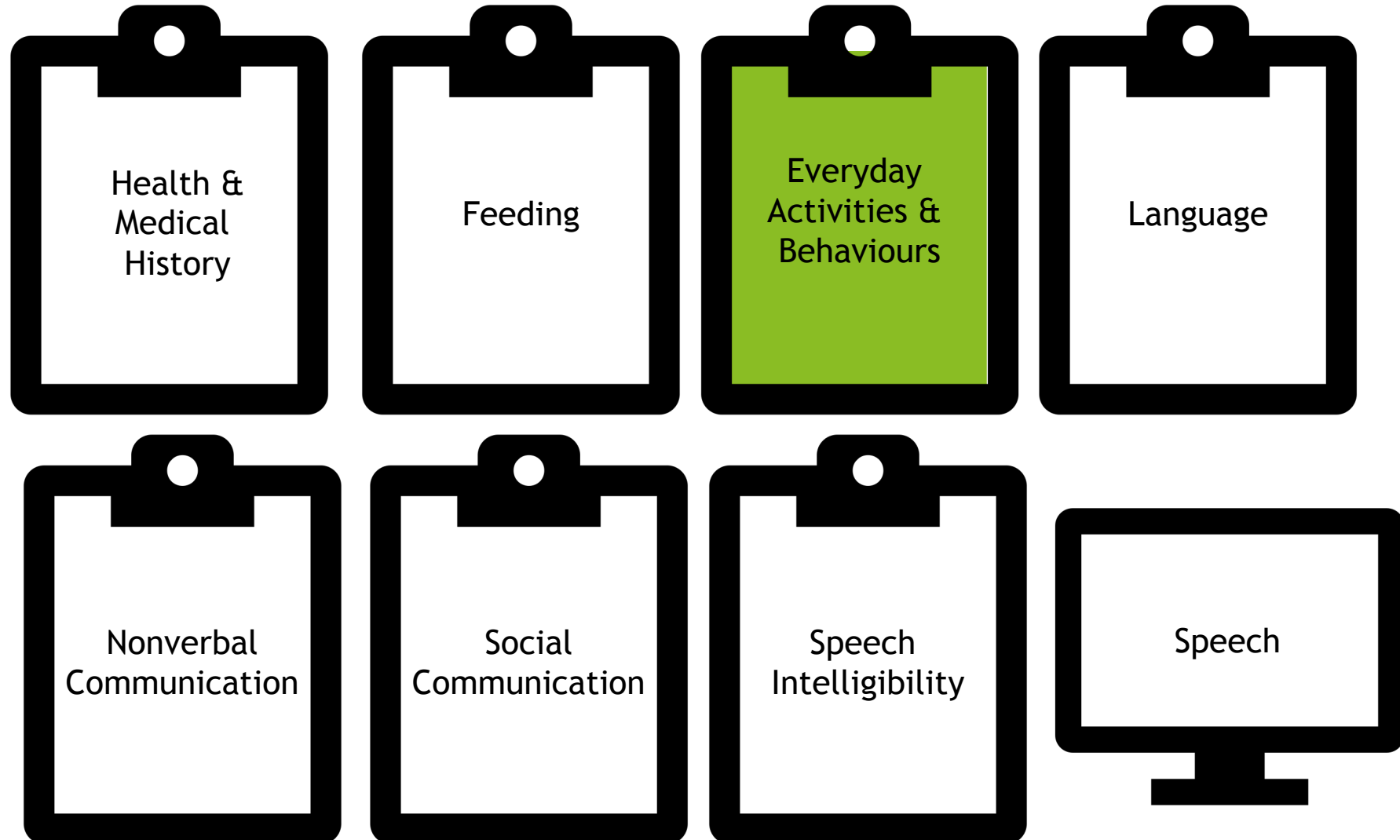
Feeding



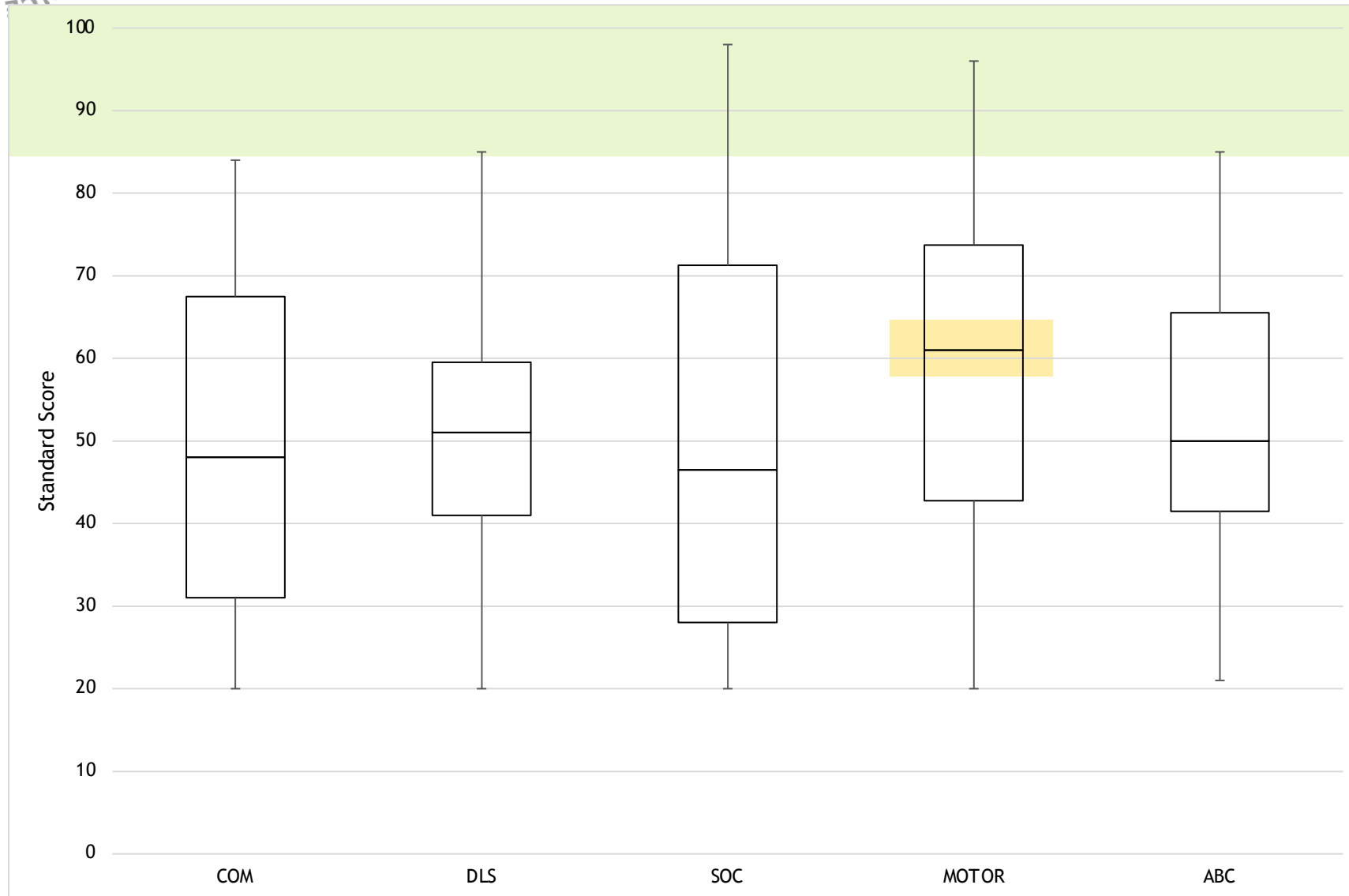
- History of feeding challenges
- Nasogastric and gastrostomy tubes
- Over stuffing, pocketing and difficulty moving food with tongue
- Oral aversion



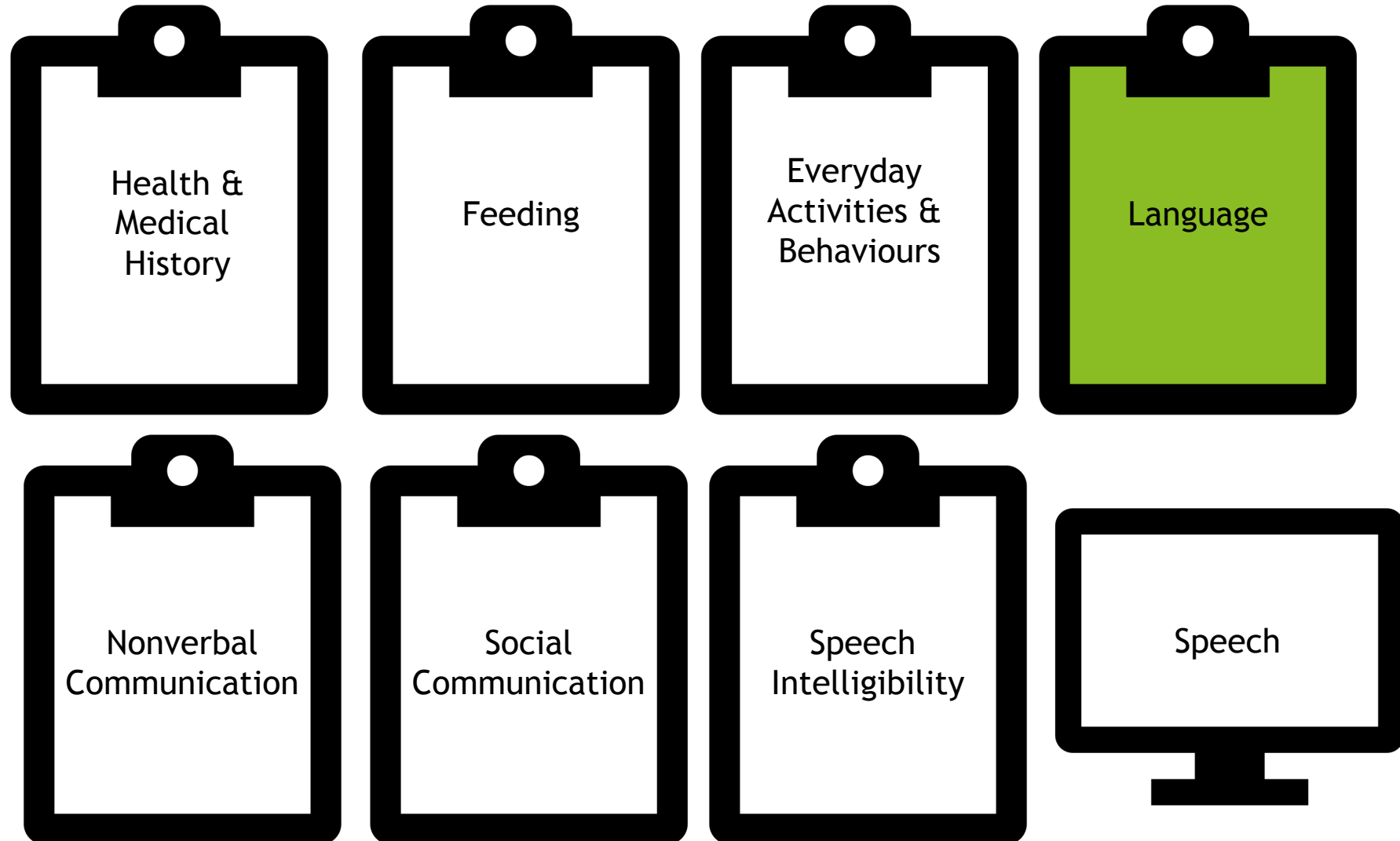
What did we assess?



Everyday Activities & Behaviours



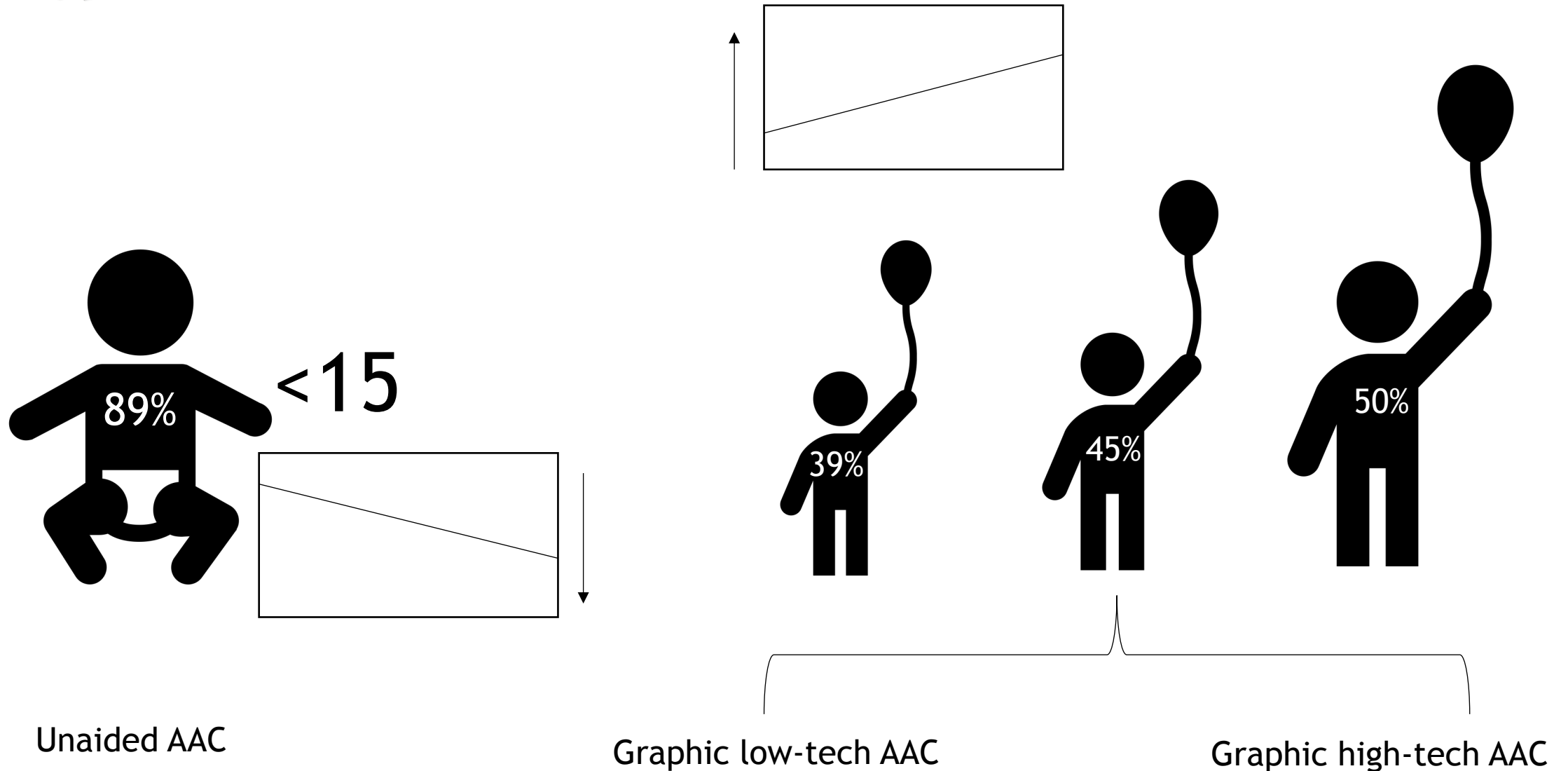
What did we assess?



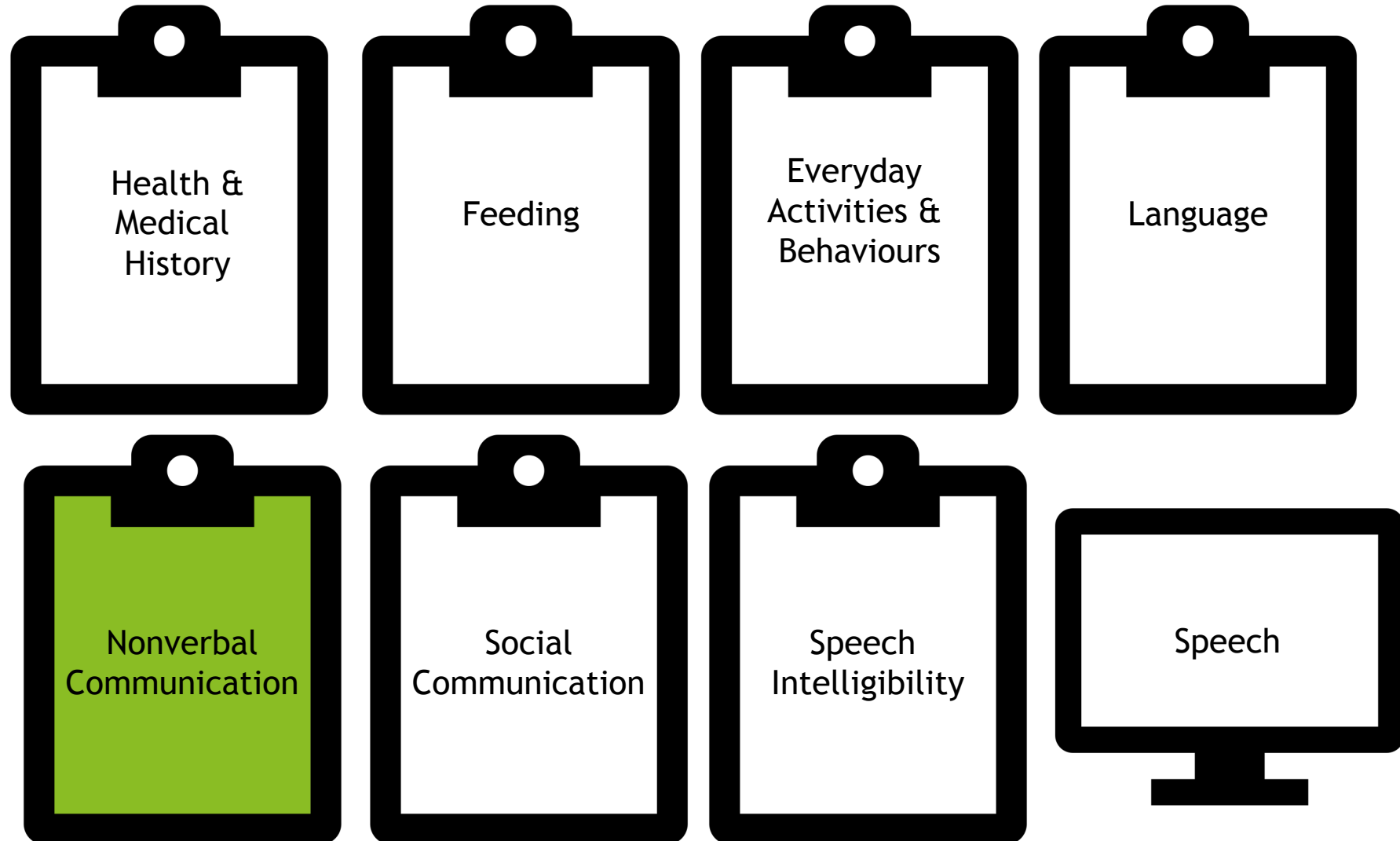
Language

- Averages across receptive, expressive, written = low
- Everyone had seen a speech pathologist/therapist
- Minimally verbal: 18/38
- Single words, short phrases: 12/38
- Conversational speech: 8/38

Augmentative & Alternative Communication



What did we assess?



	Symbolic communication (%)					Non-symbolic communication (%)					NYA*
	Speech/ words	AAC: sign language	AAC: graphic	Symbolic gesture	Facial expression	(Pre-) linguistic vocal	Non-linguistic vocal	Sterotypic behaviours	Challenging behaviours	Other non- verbal behaviours	

Social convention											
Greets others	40	20	10	15	70	55	20	40	0	70	0
Farewells others	40	25	10	25	30	40	10	5	5	60	15
Responds to name	20	5	5	20	70	15	15	5	5	55	5
Attention to self											
Gets attention	40	15	25	15	25	65	35	30	25	65	0
Seeks comfort	35	10	0	25	10	30	35	35	0	70	15
Shows off	25	0	5	15	5	25	20	5	0	35	50
Reject/protest											
Responds if routine is disrupted	25	5	10	10	5	30	60	30	35	40	60
Responds if don't like something	35	15	15	10	5	55	80	40	60	85	0
Requests object	40	15	30	35	15	60	15	0	10	55	5
Requests food	40	15	20	35	10	40	5	0	0	40	20
Requests more	35	50	15	10	5	35	10	5	0	20	15
Request action											
Requests help dressing	30	0	0	10	10	25	5	5	0	30	45
Requests the toilet	15	10	5	5	0	10	5	0	0	10	70
Requests someone to come closer	25	0	10	15	15	30	15	5	10	60	15
Request information											
Requests clarification	15	5	10	0	10	15	5	0	0	10	65
Requests information	25	10	5	5	0	15	0	0	0	10	55
Comment											
Show enjoyment	20	0	0	10	45	65	80	35	5	65	5
Show upset	25	10	5	0	25	55	70	25	30	60	0
Show boredom	10	0	5	5	55	10	10	10	10	35	40
Show amusement	20	0	5	10	30	25	90	15	0	35	5
Show fright	30	5	0	5	10	45	50	10	5	55	10
Show pain/sickness	25	5	5	20	5	15	30	15	5	35	20
Show anger	20	0	0	0	15	35	55	25	30	30	15
Show fatigue	20	5	10	0	0	20	20	15	10	45	15
Make choices											
Chooses objects	35	20	20	40	20	20	5	0	0	40	5
Chooses activities	40	20	30	45	10	25	5	0	0	30	15
Answer											
Responding yes	55	5	5	35	20	25	5	15	0	20	20
Responding no	50	5	5	25	5	25	15	0	5	15	20
Imitate											
Gesture	10	15	0	65	25	15	0	0	0	55	15
Speech	40	5	5	5	10	20	0	0	0	15	35

Speech and words

- Saying hello and goodbye
- Getting attention
- Seeking comfort
- Requesting objects and food
- Making choices
- Responding yes and no



>40%

- Sign language: requesting more
- Graphic AAC: requesting object, choosing activities
- Symbolic gesture (pointing): requesting object, food, making choices

50%

30%

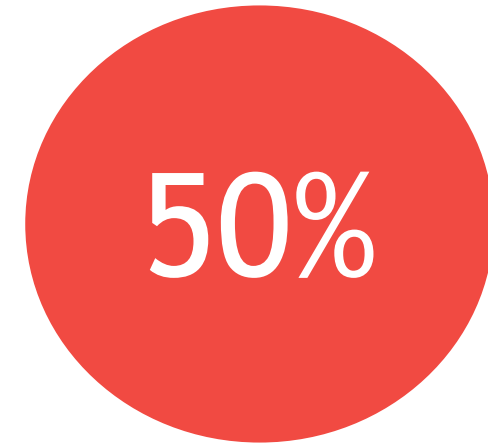
35-
40%

Non-symbolic communication

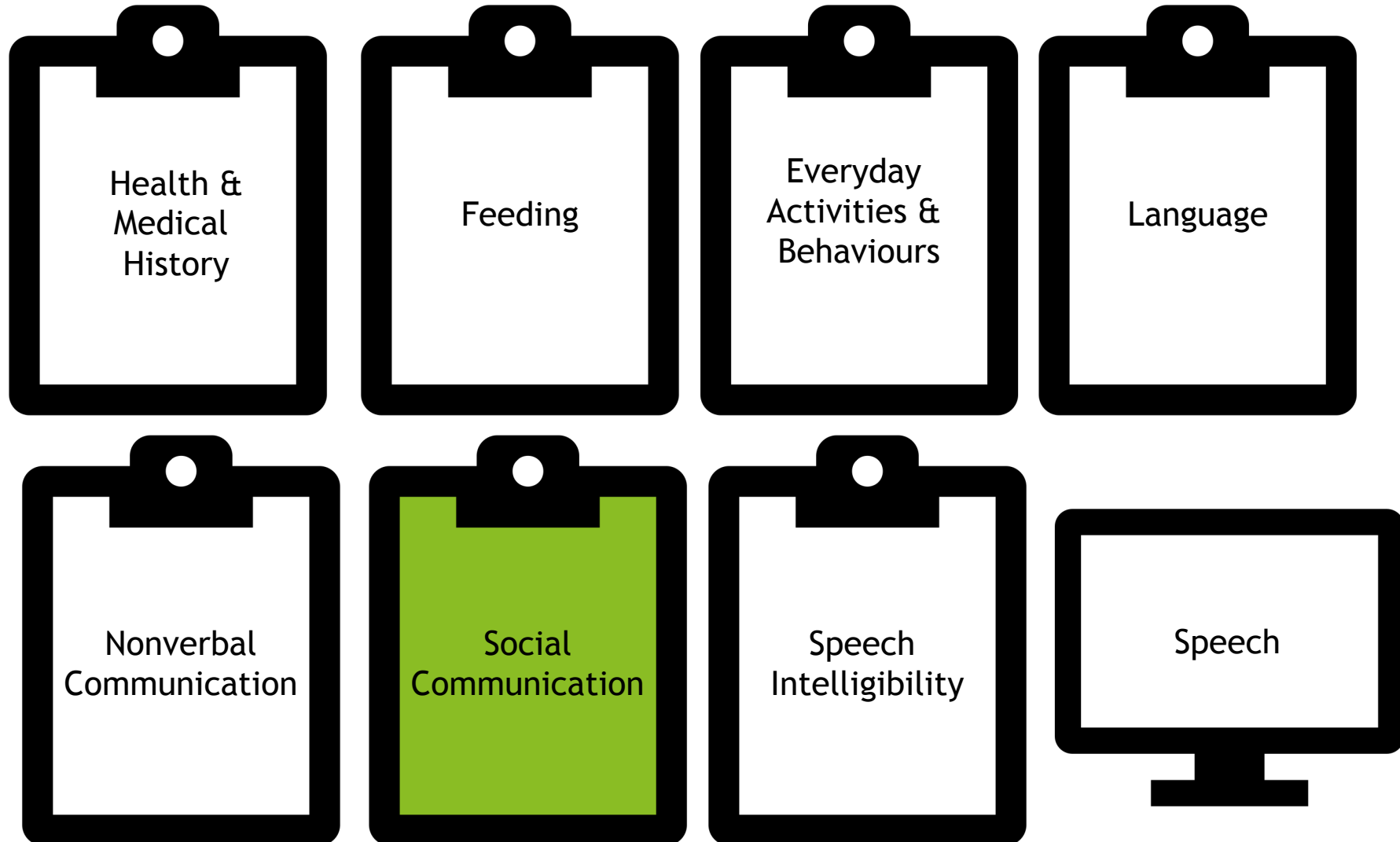
- Facial expression
- Screaming, laughing, babbling, cooing, grunting
- Stereotypic behaviours (e.g., hand flapping)
- Challenging behaviours (e.g., tantrums)

Difficulty communicating

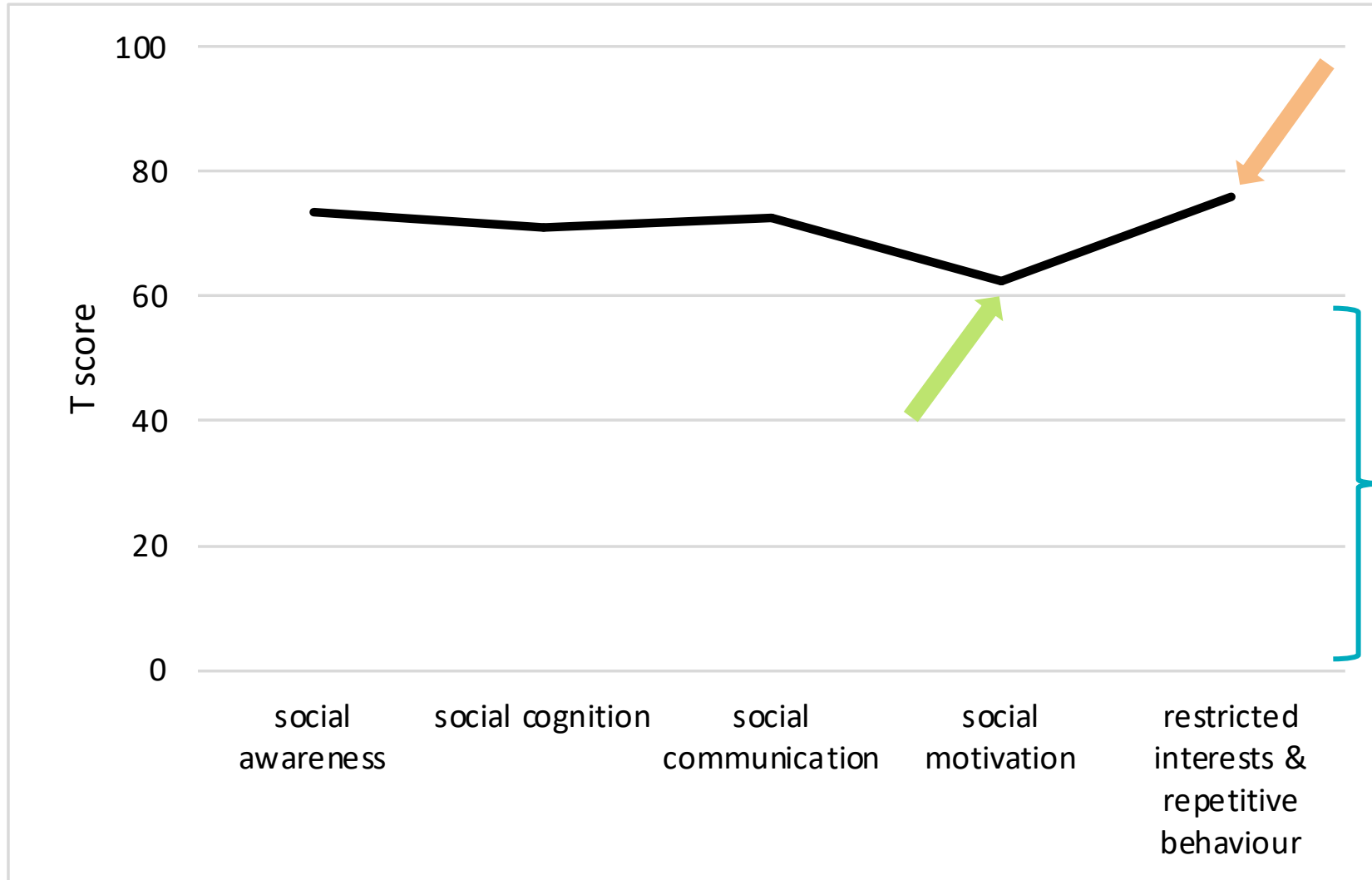
- Asking for help dressing
- Requesting to go to the toilet
- Requesting for clarification
- Requesting for information



What did we assess?



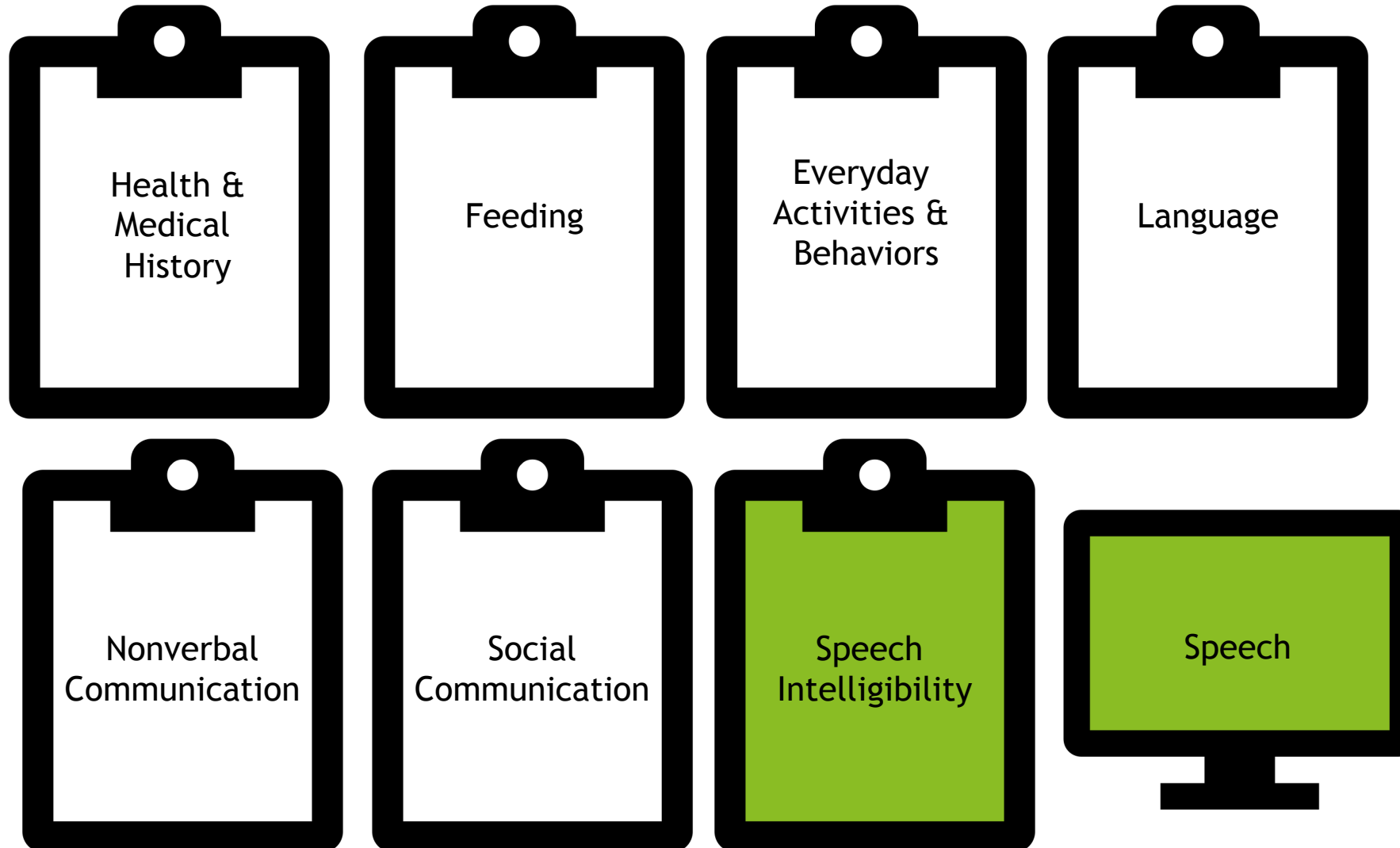
Social Communication



- Social motivation is a strength
- Restricted interests and repetitive behaviour are high

Average

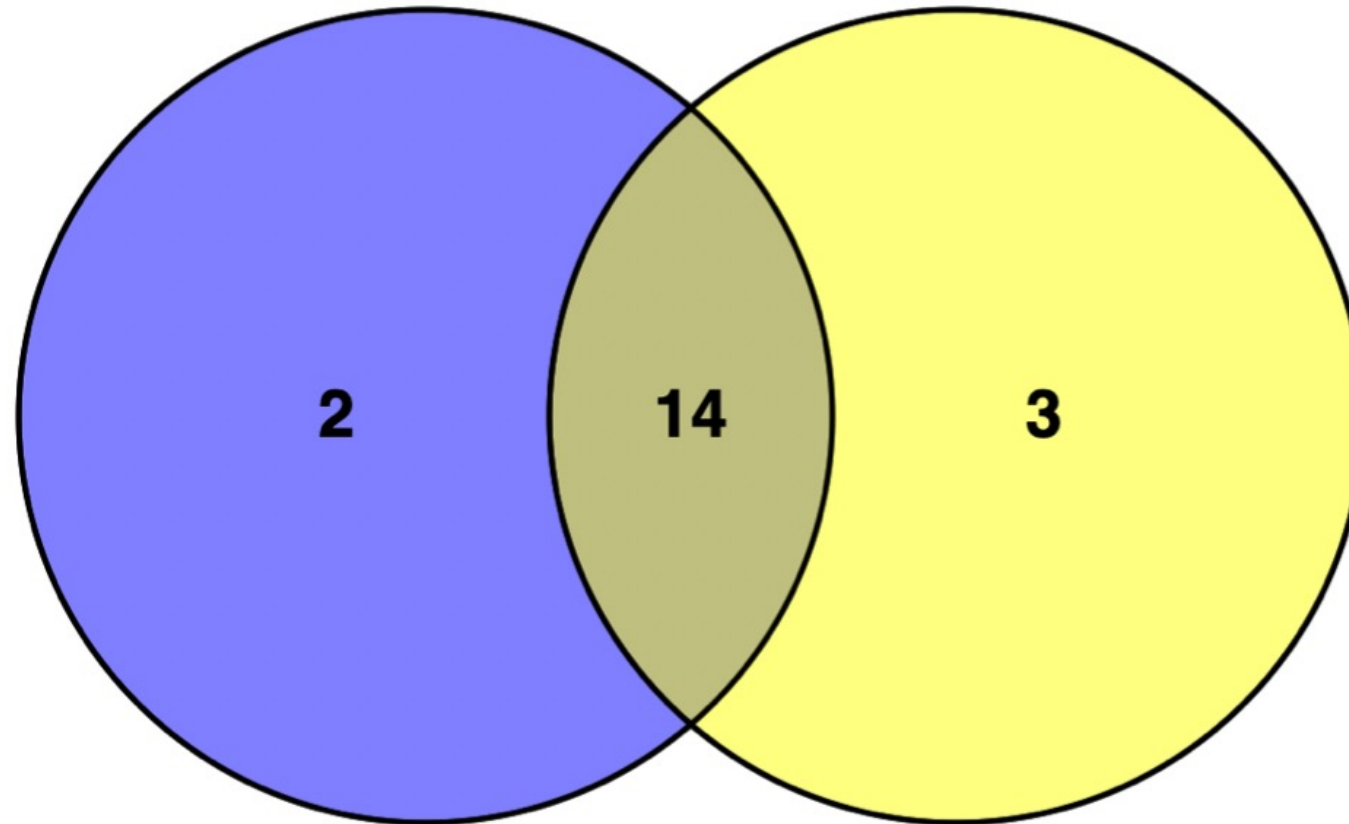
What did we assess?



Speech

Dysarthria

Apraxia



Dysarthria features

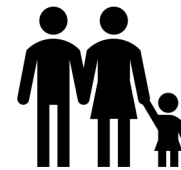


Speech intelligibility

Unfamiliar listeners



Familiar listeners



1
Never

2
Rarely

3
Sometimes

4
Usually

5
Always

What did we know now?

Speech

- Dysarthria & childhood apraxia of speech (CAS)
- Targeted therapies recommended
- Minimally verbal to conversational
- Delayed speech development

Language

- Graphic AAC recommended (+ sign language)
- Low expressive & receptive language skills
- High social motivation

What did we know now?

Feeding

- Infancy: chronic feeding impairment
- Oral aversion
- Reflux
- Oral motor impairment

Autism spectrum disorder & cognitive ability

- Comprehensive assessment recommended



Thank you

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