Persistent Speech Sound Disorder (SSD) Outcomes in a Multigenerational Family

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Conflict of Interest Disclosure

We have no financial or non financial interest or related personal interest of bias in any organization whose products or services are described, reviewed, evaluated or compared in this presentation.

Presentation outline

1. Purpose of this study
2. Overview of existing research
3. Description of this study

Purpose

Describe an idiopathic speech sound disorder (SSD) phenotype in a large nuclear family (the PM Family)

Why study this family?

- High familial aggregation of SSD
- Distributional extremes
- Large family size
- Age > 9 years

Why study this family?

Behavioural studies: Suggest a strong genetic component [a]
Molecular genetic studies: Mechanisms poorly understood, but

- families with many affected members of interest [b]
- FOXP2 gene found in single extended pedigree (KE family) [c] [d]

[a] Lewis et al., 2004; De Thorne et al., 2006.   [c] Lai et al., 2001
[b] Bailey-Wilson et al., 2011; Wijsman, 2012.   [d]Vargha-Khadem et al., 1995

Why describe the phenotype?

	language	
cognition		literacy
	speech	
phonological processing		motor
	numeracy	
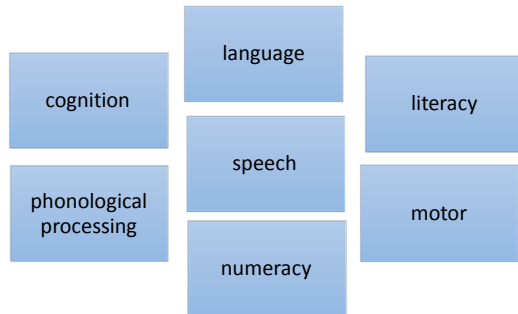
Why describe the phenotype?

May help:

- understand heterogeneity

- cross-study comparisons

- facilitate research on genetic & neural correlates

Why study persistent SSD?

- Current research focus on early childhood SSD

- In persistent SSD research

  - variable phenotypic descriptions
  - more on known than unknown origin

Persistent Speech Sound Disorder

- Persistent: Speech errors > 8-9 years of age [a]
- Speech Sound Disorder (SSD): Speech errors due to:
  structural, motor constraints, &/cognitive-linguistic constraints [b]

  Broad [c] and Narrow definitions exist [d]

([a] Shriberg, 2010; Wren et al., 2012; [b] Stein 2011; [c] ASHA; Shriberg et al., 2010, [d] DSM-5, APA, 2013)

Prevalence of SSD unknown origin:

- 15.6% of 3-year-olds   (Campbell et al., 2003)
- 3.8% of 6-year-olds   (Shriberg et al., 1999)

Prevalence of Persistent SSD: known & unknown

- 3.6% of 8-year-olds ($-1.2SD$ < mean) (Wren et al., 2012)
- 3.0% of 8-year-olds ($-2SD$ < mean)   (Wren et al., 2009)

Characteristics of persistent SSD?

Speech Characteristics

- Severity (Speake et al., 2012)
- CAS (Lewis et al., 2004b; Zaretsky et al., 2010).
- Dysarthria +/- (Fedorenko et al., 2015) (Zaretsky et al., 2010)
- Orofacial apraxia (Vargha-Khadem.. 1995)

Language & Literacy Characteristics

- Expressive language, literacy, &/phonological processing impaired [a]
- Receptive language variable [b]
- Receptive language > expressive language trend

- ([a] Lewis et al., 2004b; Speake et al., 2012; Zaretsky et al., 2010; [b] Lewis et al., 2004; Stackhouse, 1992)

Fine & Gross Limb Motor Characteristics

- Systematic assessment is rare
- Limb motor difficulties frequently been queried
  (Lewis et al., 2004b; Stackhouse & Snowling, 1992b; Zaretsky et al., 2010).

Educational/Vocational and Socio-emotional

- Little to no research specifically on persistent SSD

Study Participants

PM family: n=11

- 2 parents and 9 children
- 9 years to 55 years
- High aggregation of SSD (multiple-sound)

SSD History

SSD History	Mum	Dad	Sib 1	Sib 2	Sib 3	Sib 4	Sib 5	Sib 6	Sib 7	Sib 8	Sib 9
Sex	F	M	F	M	M	M	M	M	F	M	M
Age	51;7	55;7	28;0	24;0	20;11	16;5	17;5	15;11	13;5	10;11	9;5
SSD Grouping		Persist				Persist	Persist		Persist	Persist	Persist
History of SSD											
Received therapy for SSD											
Limited preschool Rx											
Previous CAS diagnosis											
Intelligibility at 5 years		v. poor			poor	v. poor	v. poor	poor	v. poor	v. poor	v. poor
Intelligibility at age 9 years		v. poor				v. poor	v. poor		fair	poor	v. poor
Rate of progress		v. slow				v. slow	v. slow		fair	v. slow	v. slow

Slide 1

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Participant Groups

Criteria:
- the presence of multiple-sound SSD > 9 years
- the receipt of treatment for SSD > 9 years of age

Persistent SSD group: father & siblings 4, 5, 7, 8, 9   (n = 6)
Resolved SSD group: mother & siblings 1, 2, 3, 6   (n = 5)

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Hypotheses:

(1) a core phenotype differentiated persistent from resolved SSD cases.

(2) the core phenotype resembled strongly familial persistent SSD cases in the literature.

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Assessment Protocol:
(see references in Carrigg et al 2016)

Cognition	WISC-IV, WAIS-III, + spatial working memory
Language	CELF-4, PPVT-4
Literacy & Numeracy	WIAT-II, WJ3
Speech	15 tasks -Madison Speech Assessment Protocol Conversational speech
Phonological Processing	CTOPP Nonword Repetition subtest Nonword Discrimination task Lexical Discrimination task
Oro-motor	Structure, function, & praxis tasks (MSAP)
Fine Motor	NEPSY-II: Finger Tapping, Imitating Hand Positions, Manual Motor Seq. Body praxis task

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Results

Case history & interview
- Developmental
- Academic
- Socio-emotional

Individual key results

Group comparisons

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Results: Case History - Developmental

- Non syndromal
- No comorbid developmental diagnoses
- No childhood hearing impairment
- No medical history patterns
- Primary reason for referral = speech clarity

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Case History: Academic

- All attended mainstream primary school
- Sibling 4 attended specialist high school
- Persistent cases: - Formal learning support
  - Trend to less years education

Case History: Socio-emotional

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Lasting self-consciousness re speech: (All who had treatment)

Teasing and bullying related to speech: (All persistent SSD cases)

Psychological referral recommended: (1 Resolved & 4 Persistent cases)

Severe, persisting anxiety: (Sibs 4 & 9 Social Anxiety Disorder)

"My communication disorder has had a significant and profound impact on my life. Growing up I often felt left out because I wasn't able to talk with other people, I wasn't able to tell other people my thoughts or if I needed something.

It was heartbreaking because I knew what I wanted to say, but I couldn't say it. I still feel deeply sad about not talking to others"

(Sibling 4 email using literacy support software; Carrigg et al., 2015 p. 46)

Key Individual Test Results   Shaded = affected

	Mo	Fa	Sib 1	Sib 2	Sib 3	Sib 4	Sib 5	Sib 6	Sib 7	Sib 8	Sib 9
Nonword Repetition SS	4	2	5	4	4	*	1	4	1	1	2
Nonword Repetition SS ½		2				*	1		1	1	2
Multisyllabic PPC %	92	60	95	94	84	60	66	92	73	44	19
Conversational PPC %	98.66	86.18	98.91	92.88	91.09	*	87.68	92.71	87.63	71.11	59.68
Full Scale IQ	90	97	116	108	98	75	75	103	92	85	71
Non Verbal IQ	89	101	109	99	91	106	84	106	94	79	88
Verbal IQ	96	88	124	118	107	57	79	96	83	81	69
Receptive Vocab (PPVT-4)	91	81	97	103	97	81	75	93	77	76	83
Core Language Score CELF4	97	51	109	109	96	*	61	94	78	68	65
Expressive Language (ELI)	93	49	110	108	93	*	55	95	70	55	61
Receptive Language (RLI)	97	63	*	102	102	*	71	91	88	88	92
RL>EL Gap		Y				Y	Y		Y	Y	Y
Word Reading	81	51	96	103	101	*	59	92	77	58	51
Nonword Reading	84	74	103	103	94	*	55	97	70	66	67
Word Spelling	92	50	101	92	100	65	67	84	82	59	55
Written Expression Rank	3	0	4	4	4	2	1	3	2	1	0
Nonword Discrim %	93	70	95	95	95	72	78	88	75	70	70

Cognition: Persistent v Resolved SSD Groups

	Persistent SSD			Resolved SSD					
Measure	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
Full Scale IQ	6	80	71-97	5	103	90-106	0.017*	2.39	0.75
Nonverbal IQ	6	91	79-106	5	99	89-109	0.234	1.19	0.38
Verbal IQ	6	80	57-88	5	107	96-124	0.006**	2.75	0.87
Working Memory	6	88.50	68-107	5	99	94-107	0.118	1.56	0.49
Processing Speed	6	89.50	73-104	5	96	88-103	0.521	0.64	0.20

Language: Persistent v Resolved SSD groups

	Persistent SSD			Resolved SSD					
Measure	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
PPVT-4	6	79	75-83	5	97	91-103	0.006**	2.75	0.87
Expressive Language	5	55	49-70	5	95	93-110	0.009**	2.63	0.88
Receptive Language	5	88	63-92	4	99.50	91-102	0.026*	2.22	0.79
Language Content	5	66	61-83	4	96	87-102	0.014*	2.46	0.87
Language Memory	5	68	56-78	4	92.50	85-104	0.014*	2.46	0.87

Literacy and Numeracy: Persistent v Resolved SSD Groups

	Persistent			Resolved					
Measure	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
Word Reading	5	58	51-77	5	96	81-103	0.009**	2.62	0.87
Nonword reading	5	67	55-74	5	97	84-103	0.009**	2.62	0.87
Word Spelling	6	62	50-82	5	92	84-101	0.006**	2.74	0.87
Written Expression:									
- Holistic (0-6)	6	1	0-2	5	4	3-4	0.005**	2.80	0.89
- Spelling (0-4)	6	0.00	0-2	5	1	0-2	0.364	0.91	0.29
- Punctuation (0-4)	6	0.50	0-1	5	1	1-3	0.035*	2.11	0.67
Passage Comprehension	6	74	67-85	5	96	78-108	0.022*	2.29	0.72

Literacy and Numeracy: Persistent v Resolved SSD Groups

	Persistent			Resolved					
Measure	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
Word Reading	5	58	51-77	5	96	81-103	**0.009****	2.62	0.87
Nonword reading	5	67	55-74	5	97	84-103	**0.009****	2.62	0.87
Word Spelling	6	62	50-82	5	92	84-101	**0.006****	2.74	0.87
Written Expression:									
- Holistic (0-6)	6	1	0-2	5	4	3-4	**0.005****	2.80	0.89
- Spelling (0-4)	6	0.00	0-2	5	1	0-2	0.364	0.91	0.29
- Punctuation (0-4)	6	0.50	0-1	5	1	1-3	0.035*	2.11	0.67
Passage Comprehension	6	74	67-85	5	96	78-108	0.022*	2.29	0.72

Speech & Phonological Processing: Persistent v Resolved SSD Groups

	Persistent SSD			Resolved SSD					
Measure	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
Nonword discrimination %	6	71	65-78	5	95	88-95	**0.006****	2.77	0.88
Nonword repetition SS	5	1	1-2	5	4	4-5	**0.006****	2.74	0.91
Syllable repetition %	5	58	56-86	5	84	76-96	0.057	1.90	0.63
Lexical discrimination	6	93	80-97	5	97	90-100	0.266	1.11	0.35
Conversation PPC	5	86	60-88	5	93	91-99	**0.009****	2.61	0.87
Multisyllabic words PPC	6	60.5	18-73	5	93	86-95	**0.006****	2.74	0.87
Nonword repetition PPC	5	46	36-61	5	80	63-84	**0.009****	2.61	0.87

Sibling 5, 17 years   Discussion about his speech

"Longer words. Longer words have more syllables in it and, like, I have to get them together. Because it might got a /ch/ in the middle of the thing or a double 'L' word like loon, balloon, like a big word. The bigger the word, it's harder"

"If I slow it down. But you can't slow it down when you're talking; you have to say it real fast"

Sibling 5: 17 years. Multisyllabic Words Task (MSAP)

- Emphasis
- Sympathise
- Fudgesicle
- Consciousness
- Fire extinguisher   "that's hard, pass that one"
- Statistician   "it's hard because I don't got someone talking saying it, like a computer saying, it's hard"

Father: Multisyllabic Words Task (MSAP)

- Orchestra
- Specific
- Statistics
- Fire extinguisher   "I can't say that one… I can't"
- Episcopal church

Oromotor Tasks   shaded = affected   * = reported

	Mo	Fa	Sib1	Sib2	Sib3	Sib4	Sib5	Sib6	Sib7	Sib8	Sib9
Sex	F	M	F	M	M	M	M	M	F	M	M
Oral Structure											
Orofacial Apraxia						*					
Oromotor function											
Speech-like task /z/											
Speech-like task DDK						*	?				

Motor: Persistent v Resolved Groups

Motor	Persistent			Resolved					
	n	Mdn	Range	n	Mdn	Range	p	Z	r_{spb}
Finger Tapping Repetition-SS	6	14	10-14	5	14	12-14	0.486	0.70	0.22
Finger Tapping Sequence-SS	6	10	8-11	5	11	5-12	0.299	1.04	0.33
Imitating Hand Positions SS	6	6	3-13	5	8	5-13	0.263	1.12	0.35
Manual Motor Sequences	6	2	1-4	5	4	1-4	0.153	1.43	0.45

Summary

Hypothesis 1: SUPPORTED

- a core phenotype differentiated groups
- characterised multiple Verbal Trait Disorder

Significant group differences (p < 0.01)

1. Speech accuracy (multisyllabic, nonwords, conversation)
2. Verbal IQ
3. Receptive vocabulary
4. Expressive language
5. Written expression
6. Word reading & nonword reading
7. Word spelling
8. Nonword repetition
9. Nonword discrimination

Persistent group characteristics: Core phenotype

1. Current CAS (severe → mild)
2. Severe Expressive Language Disorder
3. Impaired single word receptive vocabulary
4. Receptive-Expressive language gap (RL > EL)
5. Lower verbal IQ than resolved cases
6. Impaired reading and spelling
7. Severely impaired phonological memory
8. Impaired nonword discrimination

Persistent group: Associated characteristics

1. Academic difficulties: Formal learning support
2. Speech Intelligibility at 5 yrs: very poor
3. Speech Intelligibility at 9 years: fair → very poor
4. Progress rate: fair → very slow.

Resolved group characteristics

1. Verbal IQ and nonverbal IQ: WNL
2. Expressive and receptive language: WNL
2. Speech: WNL → minimal distortion errors
3. Literacy: overwhelmingly WNL. Vulnerabilities
4. Impaired nonword repetition

Persistent group comparison to literature

Hypothesis 2: PARTIALLY SUPPORTED

- Caution required when comparing cases
- Phenotypic similarities and differences to KE family
- More similar to published idiopathic cases

Implications for management

multiple Verbal Trait Disorder

Assessment

- multiple domains
- challenges due to ↓ unintelligibility
- multidisciplinary
- family history

Implications for management

Treatment

- multiple domains
- approach
- evaluation
- AAC
- time intensive, flexible service delivery
- multidisciplinary (motor, psychological, educational needs)

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