Linguistic and Nonverbal Abilities over Time in a Child Case of 22q11 Deletion Syndrome

Maria Kambanaros & Kleanthes K. Grohmann

The aim of this study is to profile the cognitive–linguistic performance of a male child (P.I.) with 22q11 deletion syndrome (22q11DS). Specifically, receptive and expressive language performance and nonverbal IQ (NVIQ) are described at two different time points—when P.I. was 6 and 10 years of age, respectively. Using case-based methodology, P.I.'s NVIQ and performance on global and structured language tasks are compared to typically developing children of the same chronological age and school-aged children with specific language impairment (SLI). The results show no improvement in NVIQ or vocabulary, but his morphosyntactic abilities did improve over time. The findings are discussed in relation to two hypotheses, either that the profile of language impairment in children with 22q11DS is distinctive to the syndrome or that there is co-morbidity with SLI. This is particularly important for speech–language therapists who have a primary role in diagnosing communication deficits and providing treatment.

Keywords: 22q11 deletion syndrome; co-morbidity; cognitive–linguistic profile; complex syntax; faculty of language; language development; specific language impairment

1. Introduction

In chapter 6 of *Biological Foundations of Language*, Eric Lenneberg included a short section on types of "evidence for inheritance of language potential" (Lenneberg 1967: 248ff.). Since there was not much research available at the time, it stressed the importance of family histories for establishing a genetic base of language impairments and arguably laid the foundations for a long series of twin studies research on language (see e.g. Stromswold 1998, 2001, 2006, and the relevant literature cited). The present paper can be seen as a contribution to the endeavor of detailing language and cognition under special genetic circumstances (22q11 deletion syndrome), also providing further evidence that a "pathologically lowered IQ […] does not result in bizarre use of language" (Lenneberg 1967: 326).

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The importance of genomics in speech pathology (or speech and language therapy) practice is highly recognized (ASHA 2015), yet cross-linguistic research describing language and communication abilities of children with genetically linked disorders is still in its infancy. This has a negative impact on speech pathologists' ability to provide diagnosis and guide interventions, which in turn influences educational, behavioral, and psychological outcomes for children with genetic syndromes. Likewise, there is yet a lot to be learned concerning the biological underpinnings of language informing multidisciplinary linguistics research.

This study reports on the nonverbal and language abilities of a school-aged boy genetically confirmed with 22q11 deletion syndrome (22q11DS), at two different time points in his life: at 6 and at 10 years of age. Historically 22q11DS, a neurogenetic condition, has been known by many names, including DiGeorge syndrome (DGS), Shprintzen syndrome, velocardiofacial syndrome (VCFS), conotruncalanomaly face syndrome, and CATCH22 (cardiac abnormality, abnormal face, T cell deficits, cleft palate, and hypocalcemia). The present research is the first to describe the linguistic manifestations of a language deficit associated with 22q11DS for Greek, a highly inflected, morphologically complex language. A core area of investigation will be our participant's abilities in structural language, that is, his morphosyntactic abilities and performance on more complex language tasks.

Our testing battery contains several measures for probing structural language, ranging from the comprehension of complex structures, morpho-syntactic properties, and other phenomena to the production of structurally complex clauses on a narrative retell task. But the entire testing battery goes well beyond structural language. As the first research on language abilities in 22q11DS for (Cypriot) Greek, we take a broader angle and also investigate global language and cognitive abilities, including receptive vocabulary, expressive vocabulary, and nonverbal intelligence.

Furthermore, to decipher the issue of co-morbidity with specific language impairment (SLI) reviewed below, our participant's performance on all measures is compared to two groups of children, namely a group of children with typical language development and a group with a clinical diagnosis of SLI. Taking the lead from Rice (2016), we will present the outcome of our detailed testing in a comparative conceptual framework, that is, 22q11DS vs. SLI.

2. Background on 22q11DS

22q11DS results from a submicroscopic hemizygous deletion at chromosome 22q11.2 specifically at the long arm (q) 11.2 band (Woodin et al. 2001). It is an increasingly common genetic disorder affecting at least 1 in 2,000–7,000 live births (Shprintzen 2008) and represents one of the most common known recurrent copy number condition variants (Squarcione et al., 2013). It follows an autosomal dominant inheritance pattern (a child only needs to get the abnormal gene from one parent in order to inherit the disease). However, only around 10% of cases are inherited; the majority of 22q11DS occurrences are due to random

mutation (Shprintzen 2008). The phenotype is quite varied, with close to 200 clinical features identified so far as related to abnormalities of the heart, palate, velopharyngeal mechanism, immune system, central nervous system, and brain morphology (Woodin et al. 2001).

Being a highly variable disorder, much is still not known about the contributing factors to this variability. One speculation is that variation is related to how environmental factors interact with structural and sequence variation in the genome (Squarcione et al. 2013). Consequently, each child is affected differently and the symptoms vary widely, ranging from less severe to severely affected. Due to the many different body systems that can be involved in the phenotype, a multidisciplinary approach is recommended by best practice guidelines involving fetal medicine specialists, neonatologists, pediatricians, cardiologists or cardiothoracic surgeons, immunologists, cleft surgeons, speech and language therapists, endocrinologists, clinical geneticists, and general practitioners (Allgrove et al. 2012).

Furthermore, approximately 30% of individuals with 22q11DS develop schizophrenia during adolescence/early adulthood, making this syndrome a model for the disorder (Squarcione et al. 2013). Physically, children with 22q11DS tend to have similar facial features, which may include a long and narrow face, wide-set almond-shaped eyes, a broad nasal bridge and bulbous nose tip, a small mouth, small and low set ears that are folded over at the top, and an irregular skull shape (http://www.nhs.uk/conditions/digeorge-syndrome).

In contrast with the large body of literature on the behavioral and psychiatric profiles of individuals with 22q11DS (see Scandurra et al. 2013 and relevant references within), information on the pediatric population is limited. In fact, there is evidence that 22q11DS remains largely undiagnosed in many children as an isolated speech and language disorder, or a developmental delay, in the presence of few or no physical abnormalities because the large phenotypic variation renders diagnosis more difficult (Niklasson et al. 2001). Accordingly, the median age of diagnosis is reported to be as late as six and a half years (Solot et al. 2000).

The majority of individuals with 22q11DS shows relatively mild cognitive deficits, with verbal IQ often significantly higher than performance and/or nonverbal IQ. However, there are reports of individuals with low normal intelligence (IQ 71–85) and some with an IQ in excess of 85 (Niklasson et al. 2001). Individuals with 22q11DS show relative strengths in verbal ability, rote processing, verbal memory, reading, and spelling. In contrast, weaknesses have been reported in language abilities, attention, working memory, executive functions, visuospatial memory, and psychosocial functioning (see Woodin et al. 2001 for both points).

Most significantly, research on the manifestations of speech and language disorders in children with 22q11DS is not prominent, despite communication impairment hailed as the hallmark deficit, with an estimated 90% of children having some type of speech–language deficit (Persson et al. 2006). A detailed summary of available studies describing language impairment in children with 22q11DS is presented in the Appendix.

Preschool children with 22q11DS often show smaller vocabularies, word finding deficits, reduced length of sentences, delayed use of grammatical structures, and difficulties with discourse (Persson et al. 2006). Moreover, expressive language delays are often more severe than receptive language delays. By school age, there are persistent difficulties in syntax, word finding deficits, and problems with discourse organization (Solot et al. 2000, Persson et al. 2006).

Of clinical interest is the *co-morbidity of SLI* reported so far in a large 22q11DS cohort from the USA, where 36% of children were classified as having SLI on top of 22q11DS (Solot et al. 2000). A similar finding was reported in smaller case studies involving four Dutch children with 22q11DS and SLI (Goorhuis-Brouwer et al. 2003). SLI is a term applied to children whose speech and language is substantially below age level for no apparent reason, in the absence of neurological damage, impaired sensorimotor abilities, and so on (i.e., with normal intelligence levels, hearing, vision, etc.). The reader is referred to Bishop (2014) for a more recent definition of SLI.

Directly relevant to our research is the study from Sweden involving preschoolers and school-aged children with 22q11DS (Persson et al. 2006). Here, 19 children between 5 and 8 years of age genetically diagnosed with 22q11DS (10 girls) were assessed on receptive vocabulary knowledge using the Swedish version of the Peabody Picture Vocabulary Test (PPVT; Dunn & Dunn 1981), and on narrative retell performance using the Bus Story Test (BST; Renfrew 1997). The mean full-scale IQ of the group was 78 and six children had an additional autism spectrum disorder, attention deficit/hyperactivity disorder, or a combination of the two. For the PPVT, results revealed that the majority of children (n=14) scored moderately low on receptive vocabulary, revealing more difficulties with understanding single words/concepts beyond that expected for performance or nonverbal IQ.

With regard to the BST, the majority of the 22q11DS children (n=19) scored below the mean on the information score of the task. Of clinical interest was the finding of a negative correlation between age and the information score: The older the children were, the more difficulties they had recalling information on the BST. Furthermore, all but one participant had a shorter average sentence length than expected according to chronological age norms. In contrast, five participants produced subordinate clauses within normal limits, while 14 had a lower number of subordinate clauses according to chronological age norms, revealing low grammatical abilities. The type of qualitative errors analyzed from the 22q11DS group on the BST included grammatical errors (e.g., errors of prepositions, gender, definite articles) and incomplete utterances. Based on their findings, Persson et al. (2006) concluded that the majority of the 22q11DS participants had language impairments, and difficulties were found in all language areas investigated. The authors did not classify any member of the 22q11DS group as also having SLI due to the diverse non-linguistic characteristics of the group (e.g., behavioral difficulties, including both autism spectrum and attention deficit/hyperactivity disorders). The notion of 'specific' language impairment is commonly used in the context of otherwise normal development (see Bishop 2014).

The purpose of the present study is to profile the language abilities of one male child with 22q11DS (P.I.) and compare his performance to that reported for children with typical language development (TLD) and children with a history of SLI across a battery of linguistic measures. The aims of the study are four-fold:

1. to compare P.I.'s nonverbal and language performance across all measures with pre- and primary school-aged children with TLD for each time point;
2. to compare P.I.'s nonverbal and language performance across all measures with that of children diagnosed with SLI for each time point;
3. to investigate P.I.'s morphosyntactic abilities over time;
4. to shed light on the 22q11DS±SLI debate based on the findings.

3. Methodology

3.1. Participant

Our participant, P.I., was diagnosed with 22q11DS by the head geneticist of the Makarios Hospital Genetics Clinic in Nicosia, Cyprus, using the fluorescence in situ hybridization test (FISH) after his fifth birthday (though it was not reported to us how much genetic information was lost exactly). The FISH is specially designed to look for small groups of genes that are deleted and in the case of 22q11DS shows whether the region of chromosome 22 is present. If only one copy of chromosome 22 'lights up' with fluorescent DNA dye, rather than both copies, the test is positive for 22q11 deletion.

P.I. was born from healthy, unrelated parents who are both highly educated, with university degrees in an allied health profession (mother) and information technology (father). He also has a healthy brother who is older by three years. At diagnosis, P.I. presented with no cardiac malformation but with autoimmune disorder (thyroid disorder) and growth hormone deficiency. Hearing was tested by the Makarios Hospital Audiology Clinic and reported to be within normal limits. Also, the hospital reported no positive assessment of autism spectrum disorder symptoms or any other psychiatric condition.

When first assessed by a certified speech–language pathologist (first author) at the age of 6 years, he showed facial dysmorphia in line with 22q11DS characteristics (long narrow face, almond shaped eyes, bulbous nose, small mouth, and overfolded ear helix). The oral-peripheral motor examination revealed no structural abnormalities of the speech mechanism, including the palate, but P.I. required weekly speech therapy sessions for treatment of voice quality (e.g., hypernasality) and mild misarticulations because of velopharyngeal incompetence (VPI). He also had hypocalcaemia evidenced by poor dentition and several tooth cavities. In addition, P.I. presented with motor hypotonia and a delay in development of gross motor skills. Occupational therapy was recommended to the parents to assist with gross and fine motor skills. P.I. was enrolled in the preschool education program of a public school in Nicosia and was not receiving special education services.

At the age of 10 years, he was enrolled in grade 4 in a public school in Nicosia. He was receiving special education services predominantly for mathematics. He presented with hoarseness and reduced vocal volume, typical of VPI, but generally intelligible speech. He was also receiving regular dental care. His annual medical treatment at the Cyprus Institute of Neurology and Genetics involved full blood count for cytopaenias, serum calcium, and thyroid function. He was also being monitored yearly for autoimmune disease, height, and weight. At the time of the second testing, he was undergoing psychological evaluation for aggressive behavior.

Language testing across all measures was conducted over a three-month period at age 6 years and later at age 10 years. The reader is referred to Kambanaros et al. (2018) for a full description of P.I.'s language performance at 6 years of age.

3.2. Comparative Groups

A total of 38 Greek Cypriot bilectal pre- and primary school-aged children divided into two groups served as the comparative groups, one group with TLD and one group with a clinical diagnosis of SLI. Both groups are described in detail in the original research article on diagnosing SLI in the context of Cyprus (Theodorou et al. 2016). In line with Rowe & Grohmann (2013), we consider bilectal children those whose parents are both Greek Cypriots, who were born and raised in Cyprus, and who did not spend any large amount of time outside the country, including Greece. We did not control any more specifically for balanced input or age of exposure to Cypriot Greek and Standard Modern Greek but assumed the standard path of language development laid out in our previous research, summarized most recently in Grohmann & Kambanaros (2016).

Children in the comparative groups had (i) no known history of neurological, emotional, developmental, or behavioral problems; (ii) hearing and vision adequate for test purposes after school screening at the beginning of the school year; (iii) nonverbal performance in the broad range of normal; (iv) no gross motor difficulties; (v) normal articulation; and (vi) medium–high socio-economic status. All information was obtained from the speech–language pathologists and teachers or from the children's parents. The study was approved by the Cyprus Ministry of Education and Culture through the Pedagogical Institute.

3.2.1. Typically Developing Children (5- to 6- and 7- to 8-Year-Old TLD Groups)

Ten children (4 girls) with TLD aged between 4;5 and 6;6 with a mean age of 5;8 (SD 0.6) served as the younger TLD comparative group for time point 1, when P.I. was 6 years old, and 12 children (6 girls) aged between 6;7 and 8;7 with a mean age of 7;9 (SD 0.4) served as the older TLD comparative group for time point 2, when P.I. was 10 years old. According to the classroom teacher and parent reports, all participants in the control groups were typically developing in all respects. No child was previously referred to or had received treatment by a speech pathologist.

3.2.2. Language-Impaired Children (5- to 6- and 7- to 8-Year-Old SLI Groups)

Nine children with SLI (2 girls) aged between 4;11 and 5;11 with a mean age of 5;6 (SD 0.3) served as the language-impaired younger SLI comparative group for time point 1, when P.I. was 6 years old, and seven children with SLI (4 girls) aged between 6;2 and 8;6 with a mean age of 7;6 (SD 0.9) served as the language-impaired older SLI comparative group for time point 2, when P.I. was 10 years old. Children were diagnosed with SLI by certified speech–language pathologists based on case history information, informal testing of comprehension and production abilities, analysis of spontaneous language samples, and clinical observation. SLI diagnosis was confirmed by further testing on tools used for diagnostic purposes in Cyprus (Theodorou 2013, Theodorou et al. 2014, 2016). Children with SLI were receiving speech–language therapy services by practitioners in private settings.

3.3. Socio-Economic Status

All children came from families with a medium to high socio-economic status, as measured by mothers' education level using the European Social Survey (2010) database. We compared P.I.'s mother's education level (undergraduate university degree) to the education levels of the children's mothers in all control groups. No difference was observed in her education level compared to the younger TLD group mothers' ($t_{(9)} = 0.55$, $p = 0.60$), the older TLD group mothers' ($t_{(11)} = 1.73$, $p = 0.11$), or the older SLI groups mothers' ($t_{(6)} = 1.93$, $p = 0.10$), but the education level did differ from the younger SLI group mothers' ($t_{(8)} = 2.52$, $p < 0.05$), who had a lower mean than P.I.'s mother.

3.4. Materials and Procedures

3.4.1. Nonverbal IQ (NVIQ)

P.I. was tested on the Raven's Coloured Progressive Matrices (RCPM; Raven et al. 2000), following the Greek norms of Sideridis et al. (2015), at the age of 6 years and later when he was 10 years old. Children from both the TLD and the SLI groups were tested on the RCPM separately (Theodorou et al. 2016).

3.4.2. Language Measures

All language measures administered to P.I. and the comparative groups are described in Table 1. The reader is referred to Theodorou et al. (2016) for a detailed description of the overall testing aims, methodology, and results for the TLD and SLI groups serving as the comparative groups in this research.

Measure	Domain	Source
	Linguistic	
Diagnostic Verbal Intelligence Quotient (DVIQ)* [5 subtests]	• vocabulary (naming single concepts) • comprehension of morphosyntax • production of morphosyntax • sentence repetition • comprehension of metalinguistic concepts	Stavrakaki & Tsimpli (2000)
Peabody Picture Vocabulary Test (PPVT)*	receptive vocabulary (single words)	Simos et al. (2010)
Phonetic and Phonological Articulation Test*	articulation and phonological processing	Panhellenic Association of Logopedists (1995)
Expressive Vocabulary Test (EVT)*	expressive vocabulary (single words)	Vogindroukas et al. (2009)
Clitics-in-Islands Tool (CIT)	clitic production	Varlokosta et al. (2016)
Relative Clause Task	comprehension and production of relative clauses	Theodorou & Grohmann (2013), modified from Friedmann & Novogrodsky (2004) and Novogrodsky & Friedmann (2006)
Bus Story Test (BST) [4 measurements]	• Information score • Number of subordinate clauses produced • Number of t-units (sentences) produced • MLU–word	Theodorou & Grohmann (2010) for research purposes in Greek, based on Renfrew (1997)
	Cognition	
Raven's Coloured Progressive Matrices (RCPM)*	non-verbal performance	Raven et al. (2000)

* = the measure is norm-referenced for Greek

Table 1: A description of the cognitive and linguistic measures administered to P.I. and all comparative groups.

3.4.3. Structural Language Probes

Structural language probes are those considered to tap into morphosyntactic abilities and language complexity. For this study, the comprehension and production of morphosyntax subtests of the Diagnostic Verbal Intelligence Quotient (DVIQ), the sentence repetition subtest of the DVIQ, the number of subordinate clauses produced on the Bus Story Test (BST), performance on the Clitics-in-Islands Tool (CIT), and performance on the production and comprehension of restrictive relatives are reported in the Results section.

3.5. Scoring and Analysis

For all measures, an accuracy score was calculated by summing up the number of correct responses. For all sub-categories of the DVIQ (apart from sentence repetition), a single point was given for each correct response, and no point for each incorrect one. For sentence repetition, 3 points were given for each correct response, 2 points for each response with one error, 1 point for each response with 2 errors, and no points for responses with 3 or more errors. The main statistical analysis used was the Crawford–Howell t-test (Crawford & Howell 1998), a method developed in neuropsychology for the comparison of single cases with control groups (with small sample numbers). Using this method, P.I.'s accuracy scores on the different measures were compared to the TLD and SLI groups using a two-tailed t-test.

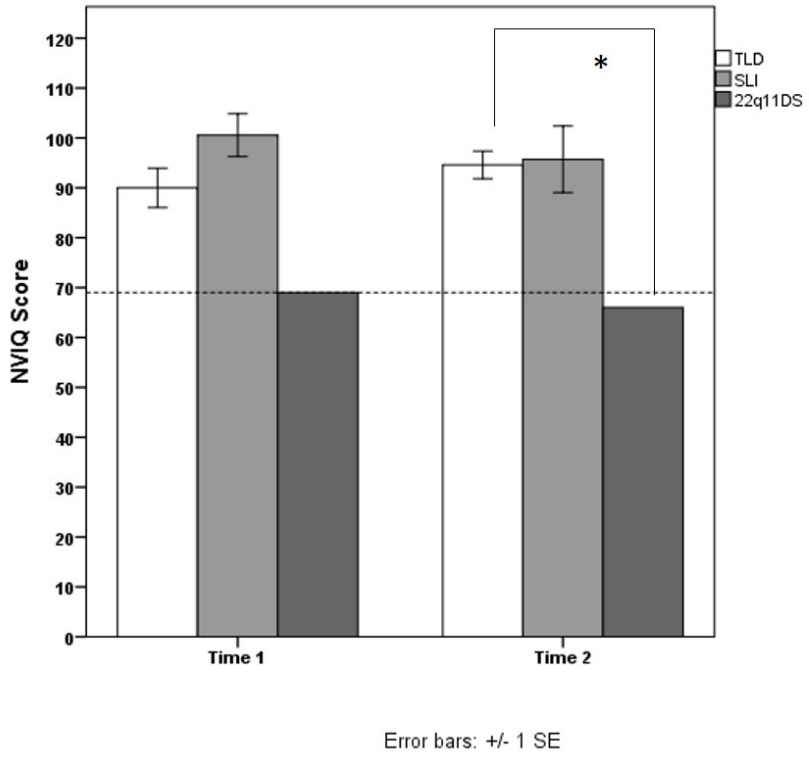
4. Results

The raw scores across all measures for P.I. and both comparative groups are reported in Table 2.

4.1. Cognition

At 6 years of age, no difference was observed between P.I.'s NVIQ and the TLD group's ($t_{(9)} = -1.61$, $p = 0.14$), and P.I. had marginally lower NVIQ than the SLI group ($t_{(8)} = -2.33$, $p = 0.05$). At 10 years of age, P.I. had lower NVIQ than the TLD ($t_{(11)} = -2.85$, $p < 0.05$) but not the SLI group ($t_{(6)} = -1.57$, $p = 0.17$). The results from the RCPM are presented in Figure 1.

Figure 1: Non-verbal IQ results (RCPM) separately for P.I. (22q11DS), TLD groups, and SLI groups at Time 1 and Time 2.



		Time 1				*Time 2*			
Measure	**Subtest (maximum score)**	**Score (SD)**			**Cut-off**	**Score (SD)**			**Cut-off**
		P.I.	***TLD–Y***	***SLI–Y***		***P.I.***	***TLD–O***	***SLI–O***	
Age		6	5;8 (0;6)	5;6 (0;3)		10	7;9 (0;6)	7;6 (0;3)	
DVIQ	Vocabulary (27)	19	22.9 (2.2)	16.8 (2.8)	19.90	20	24.7 (1.6)	20.6 (1.8)	22.35
	Production: Morphosyntax (24)	9	19.8 (2.1)	13.9 (2.7)	17.04	14	21.3 (1.4)	14.6 (1.9)	18.53
	Comprehension: Metalinguistic concepts (25)	17	19.9 (1.8)	18 (3.9)	18.73	20	22.6 (1.9)	19 (1.7)	20.14
	Comprehension: Morphosyntax (31)	16	25.4 (2.6)	24.6 (3.8)	23.85	26	28.6 (1.4)	26.4 (2.2)	26.84
	Sentence repetition (48)	35	45.5 (2.5)	40.9 (2.5)	43.18	46	47.3 (1.0)	42.3 (2.4)	44.91
	Total DVIQ Score (155)	96	133.5 (7.6)	114.1 (10.5)	124.5	126	144.5 (4.2)	122.9 (6.3)	135.0
PPVT	(212) (51)	34	63.8 (11.7)	54.8 (16.6)	56.74	23†	93.7 (25.9)	72.9 (16.7)	70.29
EVT	(50)	25	33.3 (5.1)	21.7 (2.7)	26.00	25	38.3 (3.7)	27.7 (4.8)	32.43
CIT	(12)	9	11.6 (1.3)	11 (1)	N/A	12	11.83 (0.4)	11.71 (0.5)	N/A
BST	Information	21	35.8 (11.8)	21.8 (8.9)	N/A	42	46.4 (8.9)	29.0 (8.2)	N/A
	No. of subordinate clauses produced	2	7.8 (4.1)	1.7 (1.5)	N/A	9	9 (3.0)	5.6 (1.9)	N/A
	No. of t-units (sentences) produced	10	20.6 (3.9)	15.6 (3.8)	N/A	26	20.5 (3.3)	20.1 (4.0)	N/A
	MLU–word	5.7	4.7 (1.2)	3.4 (0.7)	N/A	4.8	5.2 (1.3)	4.6 (1.1)	N/A

Key: DVIQ=Diagnostic Verbal IQ Test; PPVT=Peabody Picture Vocabulary Test; EVT=Expressive Vocabulary Test; CIT=Clitics-in-Islands Test; BST=Bus Story Test; No.=number; MLU–word=word-based mean length of utterance; 22q11DS=22q11 deletion syndrome; TLD=typical language development; Y=younger; O=older; SLI=specific language impairment; SD=standard deviation

†P.I. was tested on the shortened version of the PPVT (Simos et al., 2010) which has a maximum score of 51.

Table 2: Raw scores and standard deviations across language measures for P.I. and for all comparative groups (mean raw scores).

4.2. Global Language

4.2.1. Receptive Abilities

(i) Vocabulary (PPVT): At 6 years of age, P.I. performed similarly to both the TLD group ($t_{(9)}$ = –0.55, p = 0.59) and the SLI group ($t_{(8)}$ = –0.13, p = 0.90) for receptive vocabulary on the PPVT. P.I.'s performance at age 10 cannot be statistically compared to the comparative groups, as the shortened version of the PPVT was administered (Simos et al. 2010) and not the full battery that was used for the SLI and TLD children.

(ii) Comprehension of metalinguistic concepts (DVIQ): At 6 years of age, P.I. showed a similar performance to both the TLD group ($t_{(9)}$ = –1.54, p = 0.16) and the SLI group ($t_{(8)}$ = –0.24, p = 0.81) on the comprehension of metalinguistic concepts. At 10 years of age, P.I. again showed a similar performance to the TLD ($t_{(11)}$ = –1.32, p = 0.22) and the SLI groups ($t_{(6)}$ = 0.55, p = 0.60) on this subtest.

(iii) Information (BST): At age 6, there was a non-significant difference for this measure between P.I. and both the TLD ($t_{(9)}$ = –1.22, p = 0.25) and the SLI groups ($t_{(8)}$ = –0.08, p = 0.94). At 10 years of age, P.I. showed non-significant differences compared to the TLD ($t_{(11)}$ = –0.48, p = 0.64) and SLI groups ($t_{(6)}$ = 1.48, p = 0.19).

4.2.2. Expressive Abilities

(i) Vocabulary (EVT and DVIQ): At 6 years of age, P.I. showed a non-significant difference on the EVT compared to both groups of children, those with TLD ($t_{(9)}$ = –1.54, p = 0.16) and those with SLI ($t_{(8)}$ = 1.15, p = 0.28), and the same goes for the vocabulary production subtest of the DVIQ (TLD: $t_{(9)}$ = –1.71, p = 0.12; SLI: $t_{(8)}$ = 0.75, p = 0.48). At 10 years of age, P.I. performed significantly lower than the TLD children on the EVT ($t_{(11)}$ = –3.44, p < 0.01) and on the DVIQ ($t_{(11)}$ = –2.82, p < 0.05) but similarly to the SLI children for both the EVT ($t_{(6)}$ = –0.53, p = 0.62) and the DVIQ ($t_{(6)}$ = –0.31, p = 0.77).

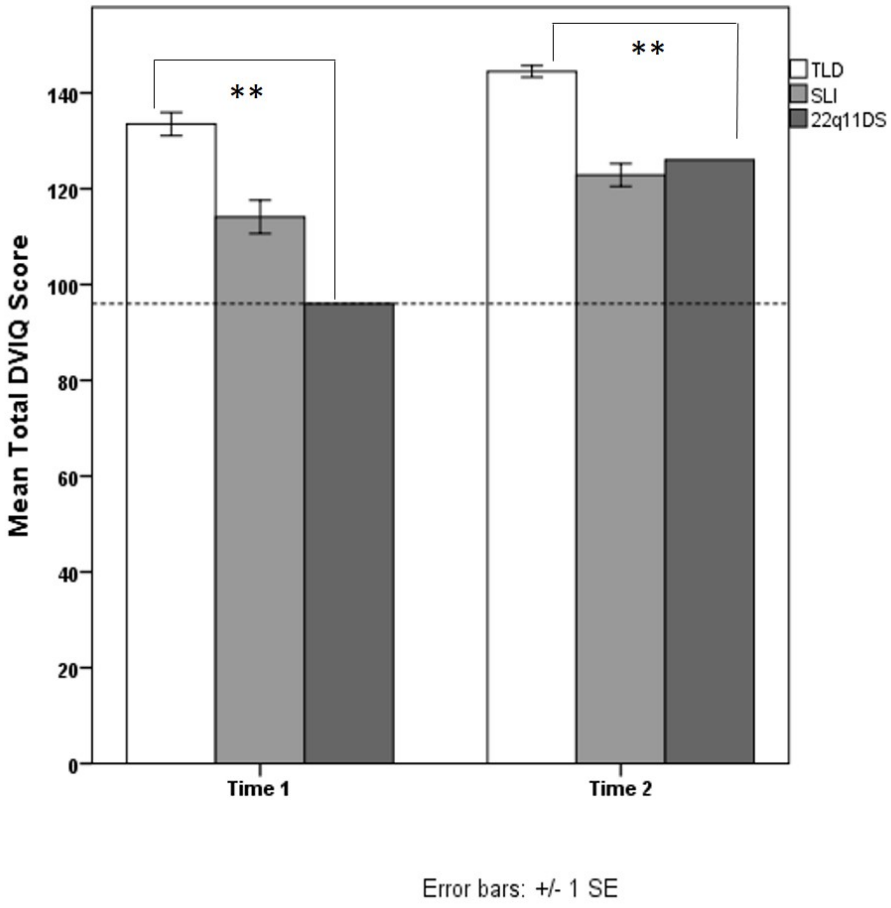
(ii) MLU–word (BST): At 6 years of age, there was a non-significant difference between P.I. and the TLD group for MLU–word ($t_{(9)}$ = –0.77, p = 0.46). When compared to the SLI group, P.I. showed higher performance ($t_{(8)}$ = 3.09, p < 0.05). At 10 years of age, however, no significant difference was observed for MLU–word between P.I. and both the TLD group ($t_{(11)}$ = –0.32, p = 0.75) and the SLI group ($t_{(6)}$ = 0.13, p = 0.90).

(iii) Number of sentences (BST): At 6 years of age, P.I. showed a significantly lower performance on the number of sentences produced measured in t-units compared to the TLD group ($t_{(9)}$ = –2.60, p < 0.05) but a non-significant performance to the SLI group ($t_{(8)}$ = –1.41, p = 0.20). At 10 years of age, P.I.'s performance was statistically similar to both the TLD group ($t_{(11)}$ = 1.59, p = 0.14) and the SLI group ($t_{(6)}$ = 1.36, p = 0.22).

4.2.3. Total DVIQ Language Score

P.I.'s total DVIQ language quotient score was significantly lower from the TLD groups at both ages (age 6: $t_{(9)} = -4.71$, $p < 0.01$; age 10: $t_{(11)} = -4.23$, $p < 0.01$). In contrast, his language performance did not differ from the SLI groups at either age (age 6: $t_{(8)} = -1.64$, $p = 0.14$; age 10: $t_{(6)} = 0.46$, $p = 0.66$). The results from the DVIQ battery are reported in Figure 2.

Figure 2: Total global language abilities (total DVIQ score) for P.I. (22q11DS), TLD groups, and SLI groups at Time 1 and Time 2.



4.3. Structural Language

(i) Morphosyntax (DVIQ subtests): At 6 years of age, P.I. differed significantly from the TLD group on the production ($t_{(9)} = -4.9$, $p < 0.001$) and comprehension of morphosyntax ($t_{(9)} = -3.46$, $p < 0.01$). On the other hand, no statistically significant difference was observed between P.I. and the SLI group ($t_{(8)} = -2.12$, $p = 0.07$ and $t_{(8)} = -1.71$, $p = 0.13$, respectively). At 10 years of age, P.I. did differ significantly from the TLD group on the production ($t_{(11)} = -5.14$, $p < 0.001$) but not on the comprehension of morphosyntax ($t_{(11)} = -1.76$, $p = 0.10$). Once more, no significant difference was observed between P.I. and the SLI group either ($t_{(6)} = -0.28$, $p = 0.79$ and $t_{(6)} = -0.18$, $p = 0.86$, respectively).

(ii) Sentence repetition (DVIQ subtest): At 6 years of age, P.I. differed significantly from the TLD ($t_{(9)}$ = –3.99, p < 0.01) but only marginally from the SLI group ($t_{(8)}$ = –2.26, p = 0.05). In contrast, at 10 years of age, he performed similarly to both the TLD group ($t_{(11)}$ = –1.29, p = 0.22) and SLI group ($t_{(6)}$ = 1.47, p = 0.19).

(iii) Number of subordinate clauses produced (BST): At 6 years of age, there was a non-significant difference for number of subordinate clauses produced between P.I. and the TLD group ($t_{(9)}$ = –1.35, p = 0.21) and between P.I. and the SLI group ($t_{(8)}$ = –0.12, p = 0.84). Also at 10 years of age, no significant difference could be observed for number of subordinate clauses produced between P.I. and the TLD group ($t_{(11)}$ = 0.00, p = 1.00) or between P.I. and the SLI group ($t_{(6)}$ = 1.69, p = 0.14).

(iv) Clitic production (CIT): At 6 years of age, P.I. showed similar performance on this task to both children with TLD ($t_{(9)}$ = –1.98, p = 0.08) and those with SLI ($t_{(8)}$ = –1.90, p = 0.09). At 10 years of age, P.I. showed similar performance on this task to both children with TLD ($t_{(11)}$ = 0.46, p = 0.66) and those with SLI ($t_{(6)}$ = 0.08, p = 0.94).

(v) Relative clauses: Comprehension and production of relative clauses was tested only when P.I. was 10 years old, given the known difficulty of the task with younger TLD children (for Cypriot Greek, using the same tool, see Theodorou & Grohmann 2013). For comprehension, P.I. performed significantly lower than both the TLD group ($t_{(11)}$ = –2.82, p < 0.05) and the SLI group ($t_{(6)}$ = –4.28, p < 0.01) on subject relative clauses, but similarly to both groups on object relatives (TLD: $t_{(11)}$ = –1.00, p = 0.34; SLI: $t_{(6)}$ = –1.34, p = 0.21). For production, no difference was observed for P.I. compared to both the TLD and the SLI groups, neither for subject (TLD: $t_{(11)}$ = 0.23, p = 0.24; SLI: $t_{(6)}$ = 1.50, p = 0.19) nor for object relative clauses (TLD: $t_{(11)}$ = 0.53, p = 0.61; SLI: $t_{(6)}$ = 1.15, p = 0.29).

4.4. Performance on Morphosyntax over Time

In total, P.I. was tested on five subtests tapping into morphosyntax (three DVIQ subtests, the CIT, and one measure from the BST narrative retell) at 6 years of age (Time 1) and at 10 years of age (Time 2). The results of the two different time points are presented in Table 3.

Structural language measure	**age 6**	**age 10**	**sign test**
DVIQ – Morphosyntax Production	9	15	+
DVIQ – Morphosyntax Comprehension	16	25	+
DVIQ – Sentence Repetition	35	46	+
CIT (production of object clitics)	9	12	+
Number of subordinate clauses produced (BST narrative retell)	2	9	+

Key: DVIQ=Diagnostic Verbal Intelligence Quotient; CIT=Clitics-in-Island-Test; BST=Bus Story Test; +=increase in scores between the two ages.

Table 3: P.I.'s performance on the structural language probes at 6 and 10 years of age, and significance reported by the sign test.

Calculations were based on the assumption that each subtest is independent of the other. The sign test, which is equivalent to the binomial test when the success probability equals 0.5, is used to explain the results. This test uses the binomial distribution to count the number of pairs *(xi, yi)* with the property *yi–xi>0* (positive sign), where *xi* denotes the score of P.I. for test *i* at Time 1 and *yi* the corresponding score at Time 2. If for a given subtest the difference is positive, this indicates improvement; if it is negative, this shows a reduction in performance. The number of morphosyntactic subtests that showed improvement at Time 2 follows the binomial distribution with five trials and success probability of 0.5. The null hypothesis—that is, no improvement in morphosyntactic ability—was tested against the alternative hypothesis, that there is indeed improvement (i.e., success probability greater than 0.5). Overall, P.I. scored higher on all morphosyntactic subtests at 10 years of age. This reveals that morphosyntactic abilities significantly improved over time ($p = 0.031, < 0.05$).

5. Discussion

The purpose of the present study was to profile, for the first time, the longitudinal trajectory of language abilities of a Greek-speaking child with 22q11DS across a number of linguistic tools used for research purposes in Cyprus. This allowed a comparison to two groups of children: one group of children with typical language development (TLD) and another with clinically diagnosed specific language impairment (SLI). Of clinical significance was the finding that for P.I., nonverbal intelligence remained stable over time.

As such, and 50 years after Lenneberg's (1967) groundbreaking work, this study may pave the way for more linguistically informed research using a developmental approach in order to understand the connection between genetically-based immuno-deficiency and cognitive–linguistic performance in 22q11DS language. The larger picture this study may allows us to paint and thereby possibly extend concerns the direction suggested by Leivada (2015). The overarching idea, most recently expanded in Leivada et al. (2017), highlights the nature and limits of language variation across child and adult populations as well as pathologies, which we address briefly below. The ultimate question underlying this approach has to do with no less than possible variation or lack thereof in the human language faculty (Tsimpli et al. 2017), a concern already detectable in Lenneberg (1967).

5.1. P.I. Compared to TLD

P.I.'s global language performance was impaired compared to TLD peers at both 6 (Time 1) and 10 years of age (Time 2), respectively, as probed by the DVIQ battery (five subtests) used for language diagnostic practice in Cyprus (Theodorou 2013, Theodorou et al. 2014, 2016). This finding lends support to the claim that language impairment is evident during both the preschool and primary school years in 22q11DS (Persson et al. 2006). In contrast, receptive language abilities (PPVT, comprehension of metalinguistic concepts, and Information score on the BST) did not significantly differentiate P.I. from TLD groups.

One possible explanation is that receptive abilities are an area of strength for P.I. (as suggested for 22q11DS by Persson et al. 2006), but this is not a commonly reported finding in 22q11DS (Glaser et al. 2002). Similarly, expressive language abilities as measured for spoken vocabulary (EVT and DVIQ Vocabulary subtest) at Time 1 were comparable to TLD peers (6 years of age) but not at Time 2, where P.I. performed significantly lower than TLD (10 years of age). In fact, P.I.'s performance on expressive vocabulary remained identical over time revealing a plateau effect.

This finding is consistent with past reports that vocabulary is a vulnerable domain in 22q11DS, and school-aged children with 22q11DS continue to struggle with word learning (Solot et al. 2000, Persson et al. 2006). Moreover, on the BST only the number of sentences produced was significantly lower for P.I. compared to TLD peers, and only at 6 years of age (Time 1), while MLU–word remained on par with TLD peers. Regarding morphosyntactic abilities as measured by the number of subordinate clauses produced on the BST, there were no significant differences between P.I. and TLD children. The BST findings in this study are not in line with what was reported for the Swedish 22q11DS cohort (Persson et al. 2006): The majority of children showed a low information score, lower number of subordinate clauses, and shorter sentence length than expected according to the Swedish BST norms.

Furthermore, in relation to structural language as measured by abilities in morphosyntax, comprehension was significantly lower for P.I. compared to TLD children at 6 years of age (Time 1) but comparable by 10 years of age (Time 2). In contrast, compared to TLD peers, production of morphosyntax was significantly impaired at 6 years of age and remained so at 10 years of age. Finally, the only other morphosyntactic measures that differentiated P.I. from TLD children were the sentence repetition subtest of the DVIQ at 6 years of age only and the comprehension of subject relative clauses in which he performed significantly worse.

The above findings corroborate earlier clinical reports that impairments in morphosyntax and complex language are robust, non-language specific features of 22q11DS (Solot et al. 2000, Goorhuis-Brower et al. 2003, Persson et al. 2006).

5.2. P.I. Compared to SLI

By comparing our participant to a group of children with SLI, that is, children with known profiles of speech and language difficulties, allows us to decipher whether the profile of 22q11DS is distinctive to the syndrome or not. Global or core language abilities (total DVIQ score) did not differentiate P.I. from the SLI groups, neither at 6 (Time 1) nor at 10 years of age (Time 2). This was also the case for receptive language abilities and for all expressive language measures—with the exception of MLU–word, where P.I. produced significantly more words than his SLI peers only at 6 years of age. In a similar vein, for structural language, P.I. performed significantly worse compared to the SLI group on sentence repetition (DVIQ subtest)—but only at 6 years of age, and only on the comprehension of subject relative clauses.

5.3. 22q11DS Morphosyntax over Time

Putting together the results of both comparative group (TLD and SLI), P.I. did perform significantly worse on two subtests from the DVIQ, production of morphosyntax and sentence repetition, and on comprehension of subject relative clauses. Unfortunately, we do not have a solid analytical knowledge base for the relevance of complex language stemming from the DVIQ subtests as markers of language difficulties. This is to say that we can describe the performance by individuals and groups, but we cannot yet pinpoint the source of deviations from the norm. Nevertheless, P.I.'s performance on the structural language probes revealed statistically significant differences over time (cf. Table 2), suggesting improvement in morphosyntactic abilities with increasing age.

This is a most encouraging finding. We can only speculate that the kind of language impairment found in 22q11DS is qualitatively different from SLI (see also right below). If so, parental language input coupled with regular schooling and specialist services do make a difference. The results suggest continued maturation of certain aspects of the language acquisition process with improvements in the 22q11DS child's language performance, even if language competence remains low.

22q11DS presents an interesting syndrome for further probing the biological underpinnings of language. One central issue concerns the invariance of the human language faculty (for recent discussion, see Tsimpli et al. 2017). According to the Locus Preservation Hypothesis (Leivada et al. 2017), operations in the computational system are not expected to be subject to impairment. This means that a 'small UG' in the sense of Hauser et al. (2002) is compatible with both invariance of the language faculty and the Locus Preservation Hypothesis, and it can address particular morphosyntactic problems in syndromes such as 22q11DS as well (see also below). This is very much work in progress, as can be witnessed from the very recent talks by Grohmann (2017) and Hinzen (2017).

5.4. The Interesting Case of Early Clitic Production

Qualitatively, there were differences between P.I., the TLD groups, and the SLI groups with clitic use probed by the CIT, a sentence completion task developed to prompt production of object clitics. The relevance of clitic production and their placement in the context of first language acquisition of Cypriot Greek has been highlighted in recent research (summarized in Grohmann 2014). P.I. behaved differently from both children with TLD and children with SLI by producing fewer clitics at 6 years of age, but this was statistically non-significant. However, he showed more omissions than either group, a phenomenon which is rare even for children with SLI (Theodorou & Grohmann 2015). Clitic production vis-à-vis omission has been taken as a clinical marker for SLI in other languages, though it is unlikely to be a clinical marker for SLI in Cypriot Greek (see Theodorou & Grohmann 2015)—and, if Leivada et al. (2017) are right, it does not tell us much about an underlying deficiency in children's language capacity either. However, at the age of 10 years, clitic production was within normal limits for P.I.

5.5. 22q11DS vs. SLI Debate

Our final aim is to tentatively use our findings to shed light on the 22q11DS±SLI debate as reported in the 22q11DS literature and outlined in the introduction. One informative approach for a more general notion of language impairment is to compare NVIQ and the linguistic performance outcomes of children with SLI to our participant with 22q11DS.

Implementing the intriguing conceptualization suggested by Rice (2016), a first comparison can be summarized as a 2×2 design with four cells of interest identified as 'A', 'B', 'C', or 'D' (as in Table 4). If P.I. has concurrent SLI (cell A), he could be compared to children with SLI who do not have 22q11DS (cell B), to children with 22q11DS who do not have SLI (cell C), and to children with TLD without either condition (cell D).

		22q11DS +	22q11DS –
SLI +		A	B
SLI –		C	D

Table 4: 2 −2 design comparison for 22q11DS and SLI (22q11DS±SLI).

At this point, we can only speculate that if A=C and A≠B, it would suggest a distinctive linguistic profile contributing to 22q11DS but not SLI. In our view, P.I.'s language-specific symptoms suggest that it is the 22q11DS variant which is the common element and that this variant is not diagnostic of SLI (cell C). This would be in line with the conclusion reached by Persson et al. (2006). Within the larger context of Lenneberg (1967), it might also contribute to a better understanding of the above-mentioned connection between genetically-based immunodeficiency and cognitive–linguistic performance in 22q11DS language.

5.6. Study Limitations

This study was a preliminary investigation of the language profile of 22q11DS compared to children with SLI (as well as typically developing control groups). While the study presents data that support further research using a comparison group of children with SLI, several limitations were apparent based on the small number of participants. Furthermore, we had no SLI child in our database with P.I.'s chronological age and home background for Time 2. This precludes big generalizations for the different populations as a whole.

However, the results of this study indicate the potential benefits of research with larger numbers of children with 22q11DS and SLI in order to tease apart the cognitive and linguistic profiles of each group. Future work will also need to focus on investigating higher-order cognitive skills like executive functions, a proven area of relative weakness according to new research in 22q11DS (Maeder et al. 2016).

5.7. Study Implications

Seen from the perspective of a larger research agenda, further exploring the exact deficits in language and cognition presented by pathologies like 22q11DS contributes to the growing research interest in *comparative biolinguistics* (Wildgen 2008, Benítez-Burraco & Boeckx 2014, Kambanaros & Grohmann 2015, among others). This program of research investigates similarities and, especially, differences in specific tasks and abilities across different pathologies, from developmental language impairments and acquired language disorders to apparently non-linguistic pathologies, that is, those that are not primarily connected to language. By so doing, we may be able to shed light on the assumed invariance of the human language faculty (cf. Grohmann 2017, Hinzen 2017, Tsimpli et al. 2017), perhaps even "uncover the locus of variation (and its constraints) across genotypes, pathologies, or across species" (Leivada 2014: 54; see the more recent Leivada et al. 2017). The present research contributes to this endeavor.

What this means is that the question of how language pathologies may inform the human language faculty in the light of Universal Grammar (UG) and vice versa receives a new twist—and it gives rise to interesting new questions (Tsimpli et al. 2017). Regardless of the outcome of these developments, UG viewed from the perspective of language pathology may open new windows into the human faculty of language as conceived today, independently of whether we assume a full-fledged faculty of language in the traditional sense ('big UG'), a highly reduced one ('small UG'), or the distinction between the faculty of language in the broad vs. narrow sense (Hauser et al. 2002)—windows that may not have been available in earlier stages of theoretically informed language research. As Tsimpli et al. (2017) put it (see also Grohmann 2017), one primary aim would be to obtain distinctive linguistic profiles regarding, say, lexical and grammatical abilities and at the same time develop cognitive profiles across a range of genetically and non-genetically different populations who are monolingual, multilingual, or somewhere in between as well as populations with or without co-morbid linguistic and/or cognitive impairments as part of their genotype.

While individual variability is clinically crucial, population-based research can advance further (cognitive–)linguistic theorizing through behavioral testing that acknowledges the brain bases involved. This will offer a unique opportunity to researchers to collaborate in fields as different as (but not restricted to) genetic biology, neurobiology of the brain, cognitive neuroscience, cognitive and developmental psychology, speech–language pathology, psycho-, neuro-, and clinical linguistics, and language development—as well as theoretical linguistics.

In addition, it may inform better about the underlying faculty(s) involved, of particular concern, of course, the role of UG in pathology. Some recent work goes in this direction, if only partially, such as emergent perspectives on autism phenotypes (Bourguignon et al. 2012), the biological nature of human language and the underlying genetic programs (Di Sciullo et al. 2010), or the idea that syntactic networks may constitute an endo-phenotype of developmental language disorders (Barceló-Coblijn et al. 2015). And if the limited research on cognitive–linguistic performance in 22q11DS reported here is on the right track, this syndrome may be very fruitful for future insights as well.

Finally, there is clinical relevance for speech pathologists to recognize the communication and language symptoms of children with 22q11DS, and to be aware of differentiating characteristics between 22q11DS, SLI, and TLD. This will facilitate improved clinical guidelines for identification and treatment of children with 22q11DS. Given the limited research regarding language function in 22q11DS to date, this is not only a first case study for (Cypriot) Greek; it also addresses larger issues of language ability in 22q11DS with respect to adaptive functioning. Overall, the findings are relevant to clinical practice by demonstrating the value of language profiling in characterizing the pattern of language impairment, with the ultimate aim of developing appropriate treatment plans.

6. Conclusion

The purpose of the present study was to provide evidence for the language profile of 22q11DS. Based on the findings of a single case, we opt for a *distinctive language profile* of 22q11DS in comparison to specific language impairment. However, further research is needed to decide on the final outcome. In that respect, we do hope that our findings provide awareness of 22q11DS. They surely constitute a first contribution to the knowledge base of the behavioral language phenotype for (Cypriot) Greek, even if only based on a single case.

There is no doubt that care of children with 22q11DS is multidisciplinary and a lifelong requirement. Early recognition is of paramount importance to improve cognitive communication skills and ultimately quality of life. Beyond that, it is very well possible that the language-based multidisciplinary research activities suggested here for the future might shed more light on the underlying questions concerning the invariance of the human language faculty across populations and syndromes (Tsimpli et al. 2017), the purported preservation of the computational system (Leivada et al. 2017), and the biological underpinnings of language today, in the 50th anniversary year of the first concrete proposals (Lenneberg 1967).

Appendix: Summary of published research on language impairment in children with 22q11DS.

Study	**Solot et al. (2000)**	**Solot et al. (2001)**	**Gerdes et al. (2001)**	**Scherer et al. (2001)**
No. of participants	305	79	112	4
Language of investigation	English	English	English	English
Age range	≤ 5	0;7–16;7	0;4–6;0	2;0–4;6
IQ range	*not provided*	For preschool children: Bayley Scales of Infant Development (BSID; *mental scale score*) 68.6 ± 13.3 and WIPSI: 84.5 ± 10.4 For school-aged children: WISC-III, VIQ: 77.8 ± 13.6, PIQ: 71.7 ± 12.8, FSIQ: 73.0 ± 12.6	For children ≥4 WPPSI-R (mean Full-scale IQ: 78 ± 12, mean Performance IQ 78 ± 12, mean Verbal IQ 81 ± 13)	BSID–2 (*mental scale score)* for VCFS (range: 50–81) and DS (range: 45–62)
Language testing	a. Preschool Language Scales–3 (PLS–3) b. Clinical Evaluation of Language Fundamentals–Revised (CELF–R) c. Goldman–Fristoe Test of Articulation d. Peabody Picture Vocabulary Test–Revised e. Expressive One Word Vocabulary Test–Revised	a. PLS–3 b. CELF–R	PLS–3, two subtests: 1. Auditory Comprehension 2. Expressive Communication	a. Sequenced Inventory of Communicative Development–Revised b. 30-minute language samples (SALT) c. CDI completed by the parents d. Speech sound analyses (MBL calculated)
Linguistic deficit	1. severe delays in expressive language in 53% of the children 2. receptive language delays in 25% of the children 3. difficulties in a variety of linguistic domains (syntax, vocabulary, concepts, word-finding, discourse organization)	1. expressive language skills significantly worse than receptive language 2. SLI pattern in 36% of the school-aged children 3. speech abnormalities of varying kinds in 75.9 % of the school-aged children	1. delayed language emergence 2. voice quality disturbances 3. low facial tone 4. articulation errors 5. dysarthria	1. fewer different words and sounds used by children with VCFS than children with DS 2. a number of different sound classes used by children with DS vs. limited sound categories used by children with VCFS 3. smaller vocabulary size for VCFS vs. DS (CDI)
Conclusion	In children with the 22q11.2 microdeletion, the emergence of language is delayed until the age of 2–3 years of age. Some children present persistent developmental delays (into school-years): These delays cannot be explained by cognitive factors, but as the **presence of specific speech and language impairment**. Presence/absence of cardiac or palatal abnormalities: no effect on development outcome.	An **SLI pattern of disorder**: Children with 22q11.2 microdeletion syndrome present (a) delayed emergence of language and (b) persistent speech and language disorders.	Speech and language delays become obvious from the first year of life in almost all children with the deletion of the 22q11.2 chromosome. These children **present a very complex developmental disorder** (including, cognitive, and language delays, as well as behaviour abnormalities).	The communicative profiles of children with VCFS vs. age-matched children with DS are different. The overall performance of children with DS is analogous to their mental-matched peers, whereas overall children with VCFS **present severe deficits** in early vocabulary acquisition and speech sound production.

Study	**Glaser et al. (2002)**	**Goorhuis-Brouwer et al. (2003)**	**Persson et al. (2006)**
No. of participants	27	4	19
Language of investigation	English	Dutch	Swedish
Age range	6;0–19;0	5;0–6;8	4;11–8;5
IQ range	Mean full scale IQ of 69.4 (SD 16.4), range: 40–105	Nonverbal IQ (normal range: 90–112)	Mean full scale IQ of 78 (range: 57–102)
Language testing	a. Receptive Vocabulary (Concept and Directions, Word Classes, Sentence Structure (6–8y.o.) or Semantic Relationships (≥9y.o.)) b. Expressive Language (Formulated Sentences, Recalling Sentences, Word Structure (6–8y.o.) or Sentence Assembly (≥9y.o.)) c. Oral Test of Word Association (letter-naming and semantic trials)	a. Language Comprehension (Dutch version of the Reynell Test of Language Development) b. Language production (Test for Sentence Development) c. Spontaneous speech sample d. Articulation and DDK rates	a. Receptive vocabulary (Swedish version of the PPVT) b. Narrative retell abilities (Swedish version of the BST) c. Articulation
Linguistic deficit	1. receptive vocabulary scores significantly lower than expressive language scores in children with VCFS, the exact opposite in the DD group 2. similar performance in the WA test for VCFS, DD, and TD groups, with semantic sores lower than the letter-naming scores	1. long sentences produced *or* 2. two- and three-word utterances produced only *or* 3. primarily nonverbal communication	1. lower scores on receptive vocabulary than expected according to NVIQ 2. difficulties in retelling a narrative (information, sentence length, number of subordinate clauses produced)
Conclusion	**Unique Developmental Patterns:** As children with VCFS get older, their expressive language skills continue to improve. MRI supports the evidence for better receptive language than expressive language skills. The weakness of both VCFS and DD is interpreted as an outcome of the general cognitive impairment.	**Children were characterized as SLI:** phonological programming deficit syndrome (2/4) or verbal dyspraxia (2/4)	**Non-SLI:** Language impairment is neutral to the issue of delay vs. disorder. The 22q11DS group had a history of recurrent otitis media and hearing loss, behavioural difficulties including ASD and ADHD, a specific behavioural phenotype, and palatal/velopharyngeal anomalies.

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