Language and Health 4 (2026) 100082

Contents lists available at ScienceDirect

Language and Health

journal homepage: www.sciencedirect.com/journal/language-and-health

Linguistic profile in individuals with 22q11.2 syndrome: A systematic review

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ARTICLE INFO

Keywords:
22q11.2 syndrome
Language
Communication
Review

ABSTRACT

22q11.2 syndrome is the second most common genetic condition worldwide, where communication, language, and speech are significantly affected to varying degrees. This has generated considerable interest among professionals working with this population. The objective of this research is to review studies conducted in recent years concerning language delays and difficulties experienced by individuals with 22q11.2 syndrome. A total of 21 articles were analyzed, focusing on the development of oral language, written language, and overall communication, following the PRISMA Statement guidelines and COSMIN methodology. The results highlight that individuals with 22q11.2 syndrome present significant difficulties, particularly in speech, various areas of expressive language, and in the acquisition of reading and writing skills when compared to their typically developing peers across different developmental stages. Therefore, linguistic intervention should be considered a fundamental area of support throughout the lifespan of individuals with 22q11.2 syndrome.

1. Introduction

22q11.2 syndrome is classified as a rare disease caused by a chromosomal alteration on chromosome 22, specifically at locus q11.2, which may involve either a deletion or duplication (Cortés-Martín et al., 2022). It is among the most common chromosomal microdeletions, with an estimated prevalence of 1 in every 2000–4000 live births, affecting both sexes equally (Cortés-Martín et al., 2022; Wierzchowski et al., 2021). In addition to being referred to as 22q11.2 deletion syndrome, this clinical entity is also known by other names, such as "DiGeorge syndrome" and "velocardiofacial syndrome." Diagnosis is based on the observation of phenotypic traits and the analysis of clinical signs and symptoms. A definitive diagnosis requires genetic confirmation (Cortés-Martín et al., 2022).

Since the first studies published in 1999, knowledge about this microdeletion has significantly advanced, allowing for a better characterization of its broad phenotypic variability. 22q11.2 syndrome is the second most frequent genetic cause of developmental delay after Down syndrome, accounting for approximately 2.4% of all cases of developmental disabilities (Wierzchowski et al., 2021).

Phenotypic manifestations in this syndrome are highly variable and may change over the course of development. Common physical traits include cardiac anomalies, gastrointestinal problems, immune deficiency, hearing loss, palatal defects, and hypocalcemia (Wierzchowski et al., 2021). From a neuropsychological perspective, individuals with 22q11.2 syndrome often present with intellectual functioning in the borderline to mild intellectual disability range from early childhood (Lorena & Sandra, 2023), typically with IQ scores between 70 and 85, although some cases may fall between 55 and 70 (Drmic et al., 2022; Gerdes et al., 1999). Beyond developmental delays, associated cognitive challenges include attentional deficits, executive functioning impairments (particularly working memory), visuospatial processing difficulties, as well as arithmetic and sensorimotor challenges (Jalal et al., 2021).

The neurocognitive profile is markedly heterogeneous both between individuals with 22q11.2 syndrome and across developmental stages. From infancy, motor delays are commonly observed, often accompanied by hypotonia, along with deficits in speech and/or language (Schneider et al., 2014). Neuropsychiatric conditions such as Attention Deficit Hyperactivity Disorder (ADHD) and Autism Spectrum Disorder (ASD) tend to emerge during early developmental stages (Morrison et al., 2020). Additionally, disorders such as anxiety and psychotic illnesses may appear at any stage of development (Zinkstok et al., 2019). Among these, schizophrenia and related psychotic disorders have been particularly studied, with a lifetime prevalence of schizophrenia estimated at around 25% (Frascarelli et al., 2020).

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https://doi.org/10.1016/j.laheal.2026.100082
Received 1 December 2025; Received in revised form 19 January 2026; Accepted 14 March 2026
Available online 21 March 2026

Speech and language impairments affect approximately 95% of children with 22q11.2 syndrome, making them one of the most frequent early childhood symptoms (Solot et al., 2019). These early language difficulties negatively impact social interactions, socioemotional development, and overall well-being. Among them, delays in expressive language stand out as one of the most commonly reported behavioral symptoms by parents (Everaert et al., 2021). Most children with 22q11.2 syndrome exhibit low speech intelligibility (Baylis & Shriberg, 2019), whose underlying causes often remain unclear and may stem from neurological conditions such as apraxia or speech sound disorders, and/or anatomical anomalies including velopharyngeal insufficiency in the absence of a cleft palate (Everaert et al., 2023).

Studies have shown that this population often presents a linguistic profile characterized by more severely impaired expressive language compared to comprehension, with difficulties articulating words, constructing grammatically correct sentences, and using age-appropriate vocabulary (Solot et al., 2019). Although receptive language abilities are generally better preserved, deficits have been reported in the comprehension of complex instructions and figurative or pragmatic language (Glaser et al., 2002; McCabe et al., 2011).

Research specifically highlights challenges in the lexical-semantic and morphosyntactic components (Solot et al., 2000; Verbesselt et al., 2023). During the school years (ages 6–12), children with 22q11.2 syndrome often exhibit difficulties in semantics, syntactic accuracy and complexity, as well as in the production and comprehension of narratives (Everaert et al., 2021). Common characteristics include limited vocabulary, word-finding difficulties, shorter sentence length, delays in grammatical structure use, and discourse-level issues (Kambanaros & Grohmann, 2017).

Linguistic impairments frequently develop into pragmatic difficulties, which negatively affect social interactions (Álvarez et al., 2009). Consequently, school-aged children with 22q11.2 syndrome often struggle to use contextual information to understand, organize, and express language appropriately, frequently focusing on secondary details (Van Den Heuvel et al., 2018). These difficulties include problems adapting to context, maintaining coherence, and understanding conversational partners. Challenges are also noted in organizing discourse and maintaining conversational turn-taking (Moraleda-Sepúlveda et al., 2023).

Non-verbal communication is also compromised, as children with 22q11.2 syndrome tend to edit information only in proximal contexts (Sebastián-Lázaro et al., 2020) and fail to accurately interpret facial expressions to infer contextual meaning (Murphy, 2004; Sebastián-Lázaro et al., 2020). Additionally, prosodic features are often affected, resulting in slowed and disrupted speech rhythm (Sebastián-Lázaro et al., 2022; Solot et al., 2000; Van Den Heuvel, Reuterskiöld et al., 2017b).

As described thus far, the scientific literature on this topic has significantly developed over the past 30 years. Therefore, the primary aim of this review is to compile the available scientific evidence regarding the development of speech, language, and communication in individuals with 22q11.2 syndrome in order to establish a linguistic profile for this population.

Given the overlap in linguistic profiles and the shared genomic region involved, studies addressing both 22q11.2 deletion and duplication provide complementary insights into the impact of this copy number variant on language and communication development. Therefore, the present systematic review includes research on both deletion and duplication cases in order to offer a comprehensive overview of the linguistic and communicative phenotype associated with 22q11.2 copy number variants.

2. Method

2.1. Search strategy

A systematic literature search was conducted in the following electronic databases: MEDLINE (via PubMed), PsycInfo, Scopus, ERIC, and Web of Science. The search aimed to identify empirical studies examining speech, language, communication, and literacy skills in individuals with 22q11.2 copy number variants, including both deletion and duplication.

The search strategy combined terms related to the genetic condition with terms referring to language and communication domains. Keywords included variations of *"22q11.2 deletion syndrome", "22q11.2 duplication", "velocardiofacial syndrome", and "DiGeorge syndrome", combined with terms such as "language", "speech", "communication", "pragmatics", "phonology", "morphosyntax", "semantics", "narrative", "reading", and "writing"*. Boolean operators (AND/OR) were used to adapt the search syntax to each database.

Searches were limited to peer-reviewed articles published in English. No initial restrictions were placed on publication year in order to capture the full range of relevant studies. In addition to the electronic database search, reference lists of the included articles were manually screened to identify further relevant publications that may not have been retrieved through the database search.

All records identified through the search process were imported into a reference management system, and duplicates were removed prior to the screening process.

2.2. Eligibility criteria

Studies were included in the present review if they met the following criteria: (1) the study population consisted of children or adolescents with a confirmed diagnosis of 22q11.2 deletion or 22q11.2 duplication; (2) the study reported original empirical data; (3) at least one outcome measure assessed speech, language, communication, or literacy-related skills, including but not limited to phonology, expressive or receptive language, semantics, morphosyntax, pragmatics, narrative abilities, reading, or writing; (4) the article was published in a peer-reviewed journal; and (5) the study was available in English.

Studies were excluded if they: (1) focused exclusively on adults; (2) did not provide specific data on speech, language, or communication outcomes; (3) consisted of reviews, meta-analyses, theoretical papers, editorials, or conference abstracts without full empirical data; or (4) reported duplicate data from previously published studies. When multiple publications reported data from the same cohort, the most comprehensive or recent study was retained.

2.3. Study selection

The initial search yielded a total of 3562 records. After removing duplicates, 90 unique records were screened based on title and abstract. Studies that appeared potentially relevant underwent full-text review to determine eligibility.

Screening and full-text assessments were conducted independently by two reviewers. Any discrepancies were resolved through discussion, and a third reviewer was consulted when consensus could not be reached.

Ultimately, 21 studies met the inclusion criteria and were included in the review. The selection process is detailed in the PRISMA flow diagram (Fig. 1), illustrating the number of studies at each stage of screening and the reasons for exclusion.

2.4. Methodological quality assessment (COSMIN)

The methodological quality of the included studies was evaluated using the **COSMIN (COnsensus-based Standards for the selection of**

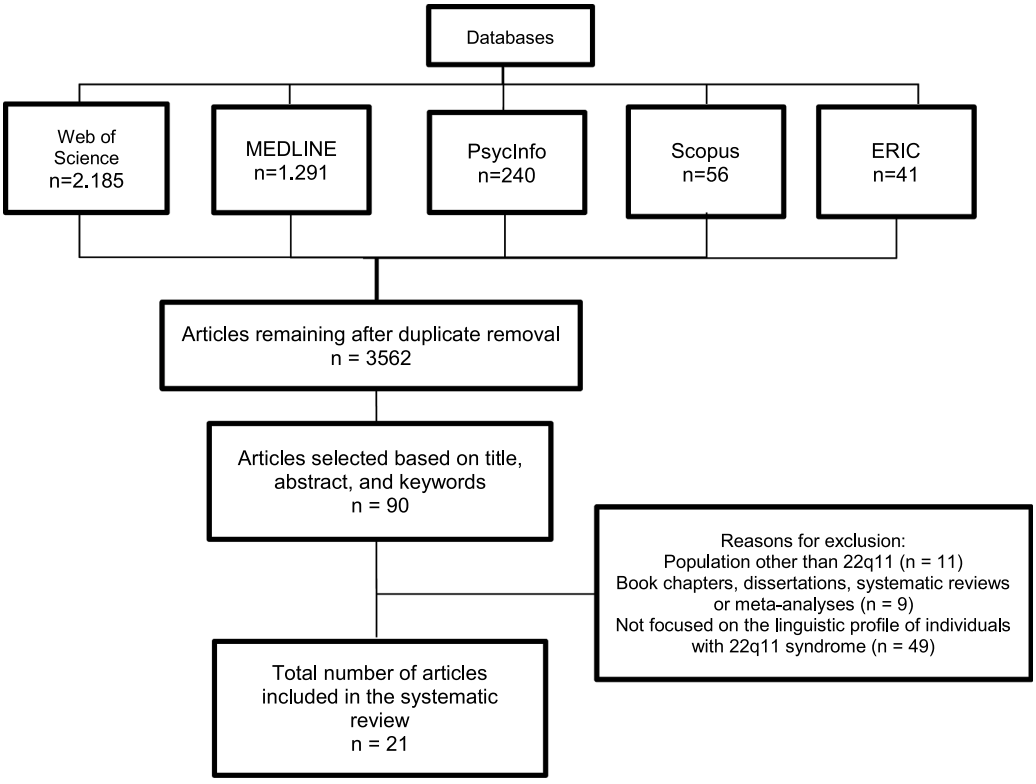

Fig. 1. Flowchart of the bibliographic selection process based on the established criteria.

health Measurement INstruments) Risk of Bias checklist, a standardized tool designed to assess the rigor of studies on measurement properties. This framework is particularly suited for evaluating instruments measuring language development and related domains.

Two reviewers independently assessed each study across key COSMIN domains, including content validity, structural validity, reliability, internal consistency, and construct validity. Ratings were assigned on a four-point scale (very good, adequate, doubtful, or inadequate), reflecting the methodological robustness of each study. Discrepancies were resolved through discussion, with a third reviewer consulted when necessary.

Applying COSMIN allowed for a systematic appraisal of the strengths and limitations of the included studies, ensuring that interpretations of language outcomes in individuals with 22q11.2 deletion syndrome are grounded in high-quality evidence. The results derived from this evaluation are detailed in Table 1.

3. Results

The analysis of the studies included in this review was conducted using the COSMIN methodology (COnsensus-based Standards for the selection of health Measurement INstruments), which allows for the systematic evaluation of the quality of the tools used in the research. This methodological approach ensures the validity and reliability of the measurements analyzed, taking into account aspects such as internal consistency, reliability, measurement error, content validity, and structural validity, among others.

Demographic and methodological variables—such as sample size, mean age of participants, sex distribution, and the components assessed in each study—were collected. These data allow for the identification of common characteristics within the studied population, providing a solid foundation for the comparison and synthesis of findings, as presented in Table 2.

The studies analyzed in the present review highlight a wide range of objectives and findings related to the linguistic profile of individuals with 22q11.2 syndrome. However, it is important to note that the assessment tools used vary considerably across studies. Table 3 summarizes the instruments employed throughout the different investigations.

Table 1
COSMIN methodology.

Investigation	A	B	C	D	E	F	G	H	I	J
Antshel et al. (2014).	-	+	-	+	+	+	-	+	+	+
Boerma et al. (2023).	-	+	-	+	-	+	-	+	+	+
Everaert et al. (2023).	+	-	+	+	-	+	+	-	-	+
Glaser et al. (2002).	+	-	+	+	?	+	?	-	-	+
Hamsho et al. (2017).	+	-	+	+	+	+	?	+	-	+
Kambanaros and Grohmann (2017).	?	+	+	+	?	+	?	-	+	+
Persson et al. (2006).	?	+	-	+	-	+	-	?	-	?
Rakonjac et al. (2024).	?	-	+	+	?	+	?	-	-	+
Rakonjac et al. (2016a).	+	+	+	+	+	+	+	+	-	-
Rakonjac et al. (2016b).	?	-	+	+	?	+	?	-	+	+
Scherer et al. (2001).	?	+	+	+	+	+	+	+	+	?
Scherer et al. (1999).	?	-	-	+	-	+	-	?	-	?
Sebastian et al. (2020).	?	-	-	+	-	+	-	?	-	?
Selten et al. (2021).	-	+	+	+	-	+	-	+	+	-
Solot et al. (2001).	?	-	-	+	-	+	-	?	-	?
Solot et al. (2000).	?	-	+	+	+	+	-	-	-	+
Van Den Heuvel et al. (2017a).	?	-	-	+	-	+	-	?	-	?
Van Den Heuvel et al. (2018).	+	-	+	+	+	+	?	-	+	+
Van Den Heuvel et al. (2017b).	?	+	-	+	-	+	-	?	-	?
Verbesselt et al. (2023).	+	-	+	+	+	+	?	-	-	+
Verbesselt et al. (2022).	+	-	+	+	+	+	?	-	-	+

Note: + positive; − negative; ? Not applicable. Box A refers to internal consistency; Box B to reliability; Box C to measurement error; Box D to content validity; Box E to structural validity; Box F to hypothesis testing; Box G to cross-cultural validity; Box H to criterion validity; Box I to responsiveness; and Box J to interpretability.

Table 2
Sociodemographic variables of the selected studies.

Authors	Sample description	Mean age (per group)	Study components
Antshel et al. (2014)	80 children with 22q11.2 deletion, 33 siblings, 40 children with ADHD and learning difficulties	11.9, 12.2, 12 years	Reading comprehension, verbal memory, working memory, executive function
Boerma et al. (2023)	Study 1: 44 children with 22q11.2 deletion, 65 with DLD, 78 with IDStudy 2: 14 children with 22q11.2 deletion, 15 with DLD	58.4, 56.7, 55.5 months104.2, 98.4 months	Expressive and receptive grammar, sentence repetition, phonological processing
Everaert et al. (2023)	44 children with 22q11.2 deletion	58.8 months	Expressive and receptive language, phonology, early lexical production
Glaser et al. (2002)	27 children with 22q11.2 deletion syndrome, 27 with idiopathic developmental delay, 54 with ID	12.4, 11.6, 11.9 years	Receptive vocabulary, expressive language, syntax, word association
Hamsho et al. (2017)	69 adolescents with 22q11.2 deletion, 50 controls	12.19, 13.02 years	Written language, spelling, syntax, executive function measures
Kambanaros & Grohmann (2017)	1 child with 22q11.2 deletion, 10 with ID, 9 with DLD	6, 5.8, 5.6 years	Phonology, syntax, expressive vocabulary, relative clause comprehension
Persson et al. (2006)	19 children with 22q11.2 deletion	7 years	Narration, phonology, syntax, receptive vocabulary, articulation
Rakonjac et al. (2024)	15 children with 22q11.2 deletion, 14 with a 22q11.2 phenotype without microdeletion, 14 with congenital heart defects, 14 controls	5.5–12 years	Expressive and receptive language, sentence production, vocabulary, articulation
Rakonjac et al. (2016a)	11 children with 22q11.2 deletion, 11 with a 22q11.2-like phenotype without microdeletion, 11 with ID	89.63, 80.22, 89 months	Articulation, oral-motor skills, phonology, expressive language
Rakonjac et al. (2016b)	2 children with 22q11.2 deletion	7.83 years	Expressive and receptive language, articulation, oral praxis
Scherer et al. (1999)	4 children with 22q11.2 deletion, 8 with ID, 8 with cleft lip and palate, 7 with isolated cleft palate	6 months (initial evaluation)	Phonology, articulation, early expressive vocabulary, oral-motor function
Scherer et al. (2001)	4 children with 22q11.2 deletion, 4 with Down syndrome	30, 38.5 months	Communicative profile, early pragmatics, gesture use, sentence production
Sebastian et al. (2020)	30 subjects with 22q11.2 deletion	12.4 years	Pragmatic language, discourse coherence, semantic fluency, narrative skills
Selten et al. (2021)	14 children with 22q11.2 deletion, 15 with DLD, 14 with ID	104.2, 98.4, 77.2 months	Narrative comprehension and production, expressive grammar, sentence complexity
Solot et al. (2001)	53 infants and 26 school-aged children with 22q11.2 deletion	7–66 months, 5.9–16.7 years	Communicative profile, early vocabulary, articulation, speech milestones
Solot et al. (2000)	305 children with 22q11.2 deletion	Childhood to adulthood	Communicative profile, reading, expressive and receptive language
Van Den Heuvel et al. (2017a)	20 children with 22q11.2 deletion, 21 with idiopathic ID, 23 with idiopathic ID and ASD	10, 9.10, 9.11 years	Sociocommunicative behaviors, pragmatic errors, context understanding
Van Den Heuvel et al. (2018)	18 children with 22q11.2 deletion, 19 with ID, 23 with ID and ASD, 30 with TD	9.8, 9.7, 9.11, 6.7 years	Expressive and receptive language, syntax, vocabulary, narrative skills
Van Den Heuvel et al. (2017b)	18 children with 22q11.2 deletion, 18 with TD, 18 younger children with TD	9.8, 9.9, 7.2 years	Referential communication skills, narrative structure, pragmatic adequacy
Verbesselt et al. (2023)	18 Dutch-speaking children with 22q11.2 duplication, 11 English-speaking children with 22q11.2 duplication; 18 Dutch-speaking children with 22q11.2 deletion, 11 English-speaking children with 22q11.2 deletion	10.10, 9.1, 10.10, 9.1 years	Morphosyntax, expressive/receptive language, vocabulary, narrative skills
Verbesselt et al. (2022)	19 children with 22q11.2 duplication, 11 unaffected siblings, 19 children with 22q11.2 deletion	10.7, 10.10, 10.7 years	Sociocommunicative skills, pragmatic language, social responsiveness

Note: VCFS – velocardiofacial syndrome, DS – Down syndrome, TD – typical development, ASD – autism spectrum disorder, DLD – developmental language disorder, ID – intellectual disability, ADHD – attention-deficit/hyperactivity disorder.

Overall, findings suggest that language impairments in 22q11.2 syndrome cannot be fully explained by general cognitive functioning. While some studies (e.g., Sebastian et al., 2020) report normal language and working memory scores, others (Rakonjac et al., 2024) indicate that expressive language deficits may exist despite typical verbal memory, highlighting domain-specific vulnerabilities.

At the speech level, various studies report specific speech impairments in this population, including oral-motor difficulties, palatal anomalies, and articulation errors (omissions, substitutions, distortions). Solot et al. (2001) found that approximately 50% of children showed persistent delays in speech milestones through school age. Certain patterns, such as velopharyngeal insufficiency, are distinctive to 22q11.2 and are not observed in non-syndromic developmental language disorder (DLD). Similarly, Rakonjac et al. (2016b) aimed to analyze the language and speech abilities of two patients with a hereditary form of 22q11.2, reporting speech development delays in both. Scherer et al. (2001) also found low speech performance in individuals with 22q11.

Table 3
Assessment instruments in the reviewed articles.

Author and year	Assessment instruments used
Antshel et al. (2014).	WISC-III, Wechsler Individual Achievement Test (WIAT-II), Gordon Diagnostic System - Continuous Performance Test (CPT), Wisconsin Card Sorting Test (WCST), Stroop Colour-Word Test, Tower of London (TOL), California Verbal Learning Test-Children's version (CVLT-C), California Verbal Learning Test (CVLT), Schedule for Affective Disorders and Schizophrenia for School-Age Children-Present and Lifetime Version (K-SADS-PL), Autism Diagnostic Interview-Revised (ADI-R) and Behaviour Assessment Scale for Children (BASC)
Boerma et al. (2023).	Clinical Evaluation of Language Fundamentals (CELF-Preschool-2-NL), Peabody Picture Vocabulary Test (PPVT-III-NL) and analysis of a game session
Everaert et al. (2023).	CELF Preschool-2-NL, PPVT-III-NL, Van Wiechen-Developmental screening instrument, analysis of spontaneous speech recordings and Cleft Audit Protocol for Speech (CAPS-A)
Glaser et al. (2002).	Parental origin DNA analysis, Clinical Evaluation of Language Fundamentals-III (CELF-III) and Oral Test of Word Association
Hamsho et al. (2017).	CPE, WCST, TOL, Visual Span Test, CVLT-C, Clinical Evaluation of Language Fundamentals (CELF- IV), K-SADS-PL, WISC-III and WIAT-II
Kambanaros and Grohmann (2017).	Raven's Coloured Progressive Matrices (RCPM), Diagnostic Verbal Intelligence Quotient (DVIQ), Peabody Picture Vocabulary Test (PPVT), Phonetic and Phonological Articulation Test, Expressive Vocabulary Test (EVT), Clitics-in-Islands Tool (CIT) and Relative Clause Task, Bus Story Test (BST)
Persson et al. (2006).	WISC-III, WPPSI, psychoacoustic test, tympanometry, Peabody Picture Vocabulary Test - Revised (PPVT-R), BST, articulation test Phonology and consonant sound inventory, repetition of sentences, spontaneous language, and velopharyngeal function tests
Rakonjac et al. (2024).	Global articulation test, story generation test, and verbal memory test.
Rakonjac et al. (2016a).	Anamnesis with data obtained from the parents, oral praxis test, global articulation test and Scale for evaluation of psychophysiological abilities of children
Rakonjac et al. (2016b).	IEPSP Test battery, story generation test, global articulation test, oral praxis test and Human Figure Drawing Test
Scherer et al. (1999).	Hearing screening, tympanometry, Bayley Scales of Infant Development-2 (BSID-2), Sequenced Inventory of Communicative Development-R (SICD), sample analysis of the language with the Systematic Analysis of Language Transcripts (SALT), Communicative Development Inventory (CDI), language sample through a video recording, Percent Consonant Correct-Revised (PCC-R) and analysis of the velopharyngeal function
Scherer et al. (2001).	BSID-2, SICD, language samples with SALT and CDI
Sebastian et al. (2020).	Ad hoc interview with the parents, Hollingshead index, CELF-4, phonological verbal fluency test FAS, Children's Communication Checklist (CCC) for parents and BST
Selten et al. (2021).	CELF-IV-NL (Dutch version), PPVT-III-NL and Multilingual Assessment Instrument for Narratives (MAIN)
Solot et al. (2001).	BSID-2, Wechsler Preschool and Primary Scales of Intelligence-Revised, WISC-III, Pre-School Language Scale-3 (PLS), Clinical Evaluation of Language Fundamental-Revised (CELF-R) and Goldman Fristoe Test of Articulation
Solot et al. (2000).	Pre-School Language Scale-3, CELF-R, Goldman-Fristoe Test of Articulation, PPVT-R, Expressive One Word Vocabulary Test-Revised, perceptual assessment scales for evaluating voice and resonance, nasometry, air flow measurements, multiview videofluoroscopy, nasoendoscopy, or a combination of these and audiometries
Van Den Heuvel et al. (2017a).	WISC-III-NL (Dutch Edition), Wechsler Preschool and Primary Scale of Intelligence-Third Edition-Dutch Edition (WPPSI-III-NL), Snijders-Oomen Non-Verbal Intelligence Test Revised age 6–40 (SON R6–40), CELF-P2-NL, CELF-4-NL, CHC model and Children's Communication Checklist-Second Edition-Dutch adaptation (CCC-2-NL)
Van Den Heuvel et al. (2018).	Video recordings and transcripts, PPVT-III-NL, CELF- IV - NL and CELF-Preschool-2-NL
Van Den Heuvel et al. (2017b).	Action Picture Test (APT) - Dutch adaptation y Dialect Sensitive Language Test (DSLT)
Verbesselt et al. (2023).	CELF- IV-NL, CELF-P2-NL y CELF-Third, Fourth and Fifth editions (CELF-III, CELF-IV y CELF-V)
Verbesselt et al. (2022).	Children's Communication Checklist-Second version (CCC-2), Social Responsiveness Scales (SRS) and Social Responsiveness Scales-Second edition (SRS-2)

Findings by Rakonjac et al. (2016a) highlight specific difficulties in speech abilities compared to control subjects without microdeletion (even though those subjects also had delayed speech development). Additionally, Rakonjac et al. (2024) identified marked articulation deficits in children with 22q11.2, emphasizing that these impairments significantly impact speech intelligibility.

Generally, in relation to language, Scherer et al. (1999) described an early onset impairment in both expressive and receptive language, which tends to worsen over time. Speech and language impairments typically appear before age 3 and persist into school age. Over time, expressive deficits become more pronounced relative to receptive abilities, resulting in increasing difficulties in articulation, sentence formation, and discourse production. Rakonjac et al. (2016a) confirmed that individuals with 22q11.2 perform at lower levels in language skills compared to control subjects without the microdeletion. Likewise, patients with a hereditary form of 22q11.2 studied by Rakonjac et al. (2016b) showed language development delays, despite no differences in the number of words or clauses produced spontaneously; and the subjects studied by Solot et al. (2001) exhibited delays in the emergence of language milestones, with persistent disorders up to school age in half of the cases.

Regarding receptive language, Glaser et al. (2002) reported that participants aged 4–12 years with VCFS exhibited lower receptive language scores compared to expressive scores and IQ-matched controls. Age appeared to moderate this pattern: in early childhood, receptive language was relatively preserved, whereas deficits in expressive language became more pronounced during school age (Boerma et al., 2023; Persson et al., 2006).

Van Den Heuvel et al. (2018) described that receptive language, which may show relatively better performance in early stages, tends to stagnate or deteriorate over time. Likewise, Scherer et al. (2001) reported that comprehension performance typically falls at or below the child's mental age.

Conversely, Scherer et al. (1999) found that deterioration in the expressive component is more pronounced than in the receptive one, a finding also supported by Rakonjac et al. (2024). These authors noted that individuals with 22q11.2 show clear impairments in expressive and articulatory skills, but not in receptive abilities. Everaert et al. (2023) also emphasized that language difficulties are more pronounced in the expressive domain.

In the phonetic-phonological domain, individuals with 22q11.2 present multiple difficulties. Persson et al. (2006) reported that only half of the participants evaluated had a complete consonant inventory. In a study during early developmental stages, Scherer et al. (1999) documented limited phonological inventories, with development far below age expectations. Additionally, findings by Scherer et al. (2001) show that individuals with VCFS produced fewer sound types than expected for their cognitive and linguistic age (and fewer than those with Down syndrome). These difficulties are compounded by findings from Rakonjac et al. (2016b), who observed distortions, substitutions, and oral praxis-related complications.

Lexical difficulties in children with 22q11.2 syndrome have also

been described by Scherer et al. (1999), who reported severe vocabulary development limitations from early ages, with a lexicon well below age expectations. Likewise, Everaert et al. (2023) found delayed emergence of first words and reduced expressive vocabulary in this population, suggesting persistent lexical development impairments (Scherer et al., 2001). Persson et al. (2006) reported limited receptive vocabulary, affecting sentence formulation and narrative production, which also compromises verbal expression efficacy. In the school context, problems have been reported in word retrieval and organization of words and phrases in narratives, affecting discourse cohesion and clarity (Solot et al., 2000). However, there is no clear consensus in this area, as Selten et al. (2021) argue that this population may exhibit vocabulary levels similar to those of younger typically developing children.

Concerning the semantic component, Sebastián et al. (2020) observed impairments in semantic fluency tasks. Solot et al. (2000) also described semantic difficulties during school age, manifested in concept comprehension, content integration, and ability to construct grammatically complex sentences based on semantic knowledge, all of which negatively affect discourse. On the contrary, Glaser et al. (2002) did not detect differences in word association tasks between the evaluated groups (children with developmental delays). Other authors point out that semantic difficulties are more severe in deletion cases compared to duplication (Verbesselt et al., 2023).

Morphosyntactic difficulties have been consistently described as part of the linguistic profile of children with 22q11.2. Kambanaros and Grohmann (2017) observed developmental progress in the use of morphosyntactic structures, although significant differences persisted compared to typically developing peers. In a more recent study, Everaert et al. (2023) identified morphosyntax as one of the most affected linguistic components, closely related to low expressive language performance and delayed emergence of first sentences. It appears that the utterances produced are short and simple, with few grammatical errors (Persson et al., 2006).

Morphosyntactic deficits commonly include omission of grammatical morphemes, simplified sentence structures, and fewer subordinate clauses. These impairments are more pronounced in deletion cases compared to duplication (Verbesselt et al., 2023) and are observed across ages 5–12 years. Children with 22q11.2 consistently perform below age expectations and below peers with typical development or DLD in expressive grammar tasks (Boerma et al., 2023; Selten et al., 2021).

Children with 22q11.2 syndrome frequently exhibit pragmatic impairments that affect the functional use of language in social contexts. In this review, "communication patterns" are defined as the ability to manage turn-taking, maintain topics, produce coherent discourse, comprehend and produce narratives, and use appropriate pragmatic cues according to context.

Several studies provide detailed examples of these difficulties. Van Den Heuvel et al. (2017a) observed frequent errors in pragmatic language, such as inappropriate responses to conversational prompts, difficulty interpreting contextual information, perseveration on secondary ideas, and vague word choices. Narrative production was often characterized by short, simple sentences with limited structural elaboration, reducing discourse cohesion and coherence (Persson et al., 2006; Van Den Heuvel et al., 2017b). Similarly, Selten et al. (2021) reported that narrative comprehension was significantly lower than in typically developing peers, though performance was comparable to children with developmental language disorder.

Semantic-pragmatic difficulties were also observed, affecting the clarity and efficiency of communication. For example, Solot et al. (2001) described persistent communication difficulties from infancy through school age, with limitations in conveying intentions and organizing discourse. Verbesselt et al. (2022) highlighted differences between deletion and duplication cases: sociocommunicative deficits were less frequent and less severe in duplication, with more varied profiles and fewer repetitive behaviors or restricted interests.

Overall, these findings indicate that pragmatic impairments in 22q11.2 syndrome are common, persistent, and affect everyday communication, independently of general language or cognitive skills, and should be carefully assessed in both clinical and research contexts.

Reading comprehension and written expression are frequently affected, likely related to deficits in vocabulary, morphosyntax, working memory, and executive function. For example, Solot et al. (2000) observed below-average reading comprehension, while Antshel et al. (2014) identified early language ability and working memory as predictors of later reading skills. Their results showed below-average reading comprehension scores (with scores decreasing over time) and word reading scores (which remained stable longitudinally). Additionally, they found that predictors for reading comprehension were associated with decoding skills, interference control, self-monitoring, and working memory. Conversely, Hamsho et al. (2017) aimed to investigate childhood predictors of written expression skills in individuals with 22q11.2. Their results indicated that executive function and early language ability were predictors in the control group. Gender also emerged as a predictor in the study's third measurement, with better outcomes in females.

4. DISCUSSION

This systematic review synthesizes current evidence on language development in individuals with 22q11.2 syndrome, highlighting the breadth and persistence of impairments across speech, morphosyntax, semantics, pragmatics, and literacy. Overall, the studies included in this review consistently demonstrate that language difficulties emerge early, affect multiple domains, and persist into school age and adolescence, although the severity and manifestation of these difficulties vary according to participant age, domain assessed, and methodological quality.

Speech and phonology emerge as areas of early and pronounced difficulty. Delays in speech onset, limited and atypical phoneme repertoires, and articulation errors were commonly reported across studies (Persson et al., 2006; Rakonjac et al., 2016a, 2016b; Scherer et al., 1999, 2001; Solot et al., 2001). Structural anomalies, such as velopharyngeal insufficiency and submucous cleft palate, along with motor planning deficits consistent with childhood apraxia of speech, contribute to reduced intelligibility and persistent articulation difficulties. These findings are highly consistent across studies with robust methodology, supporting the conclusion that early and targeted speech intervention is essential for improving both intelligibility and motor coordination.

Morphosyntactic development is similarly affected, with individuals exhibiting simplified sentence structures, frequent omissions of grammatical morphemes, and limited use of complex clauses (Persson et al., 2006; Boerma et al., 2023; Selten et al., 2021; Verbesselt et al., 2023). These impairments appear to reflect deficits in the automation of grammatical rules rather than a lack of grammatical knowledge, and they are closely associated with limitations in verbal working memory and the processing of hierarchically complex structures. While expressive morphosyntax tends to be more impaired than receptive abilities, some studies report stagnation or deterioration of comprehension over time (Van Den Heuvel et al., 2018). Notably, differences between deletion and duplication cases remain underexplored, but existing data suggest more severe deficits in deletion cases.

Vocabulary and semantic abilities are also consistently compromised. Children with 22q11.2 syndrome show reduced expressive and receptive vocabulary, difficulties in word retrieval, and limitations in processing abstract or context-dependent terms (Persson et al., 2006; Everaert et al., 2023; Selten et al., 2021). These deficits not only constrain lexical access but also affect discourse organization and communicative efficiency, with more pronounced challenges observed in tasks requiring flexible semantic integration. Although methodological heterogeneity and age differences contribute to variability across studies, the overall pattern indicates persistent and functionally

significant semantic impairments.

Pragmatic and broader communicative abilities are particularly vulnerable and have notable implications for everyday functioning. Children with 22q11.2 syndrome frequently demonstrate difficulties in maintaining discourse coherence, understanding and producing narratives, turn-taking, and interpreting social and contextual cues (Persson et al., 2006; Selten et al., 2021; Van Den Heuvel et al., 2017a, 2017b; Solot et al., 2001; Verbesselt et al., 2022). These deficits occur independently of general language ability, limiting social interaction and academic participation. Differences between deletion and duplication cases suggest slightly milder sociocommunicative impairments in duplication, though further comparative studies are needed.

Reading and writing abilities, although less frequently studied, appear similarly affected, particularly in reading comprehension and written expression (Antshel et al., 2014; Hamsho et al., 2017; Solot et al., 2000). In contrast, decoding and orthographic skills seem relatively preserved. The scarcity of studies in this domain and the use of diverse assessment tools limit the certainty of these findings, but they nonetheless indicate a need for early and individualized educational support.

The methodological quality of the reviewed studies, as assessed by the COSMIN framework, varied considerably. While studies on expressive and receptive language often employed validated instruments and robust designs, assessments of literacy and pragmatic skills were frequently based on small samples or non-standardized measures. This heterogeneity introduces risk of bias and reduces confidence in some findings, particularly in less-studied domains such as literacy and pragmatic language.

Several research gaps emerge from this synthesis. Longitudinal trajectories of language development remain insufficiently characterized, particularly for pragmatic skills and literacy. Comparative studies between deletion and duplication cases are scarce, limiting conclusions about differential severity. Few studies integrate functional outcomes, such as social participation or academic achievement, with linguistic profiles. The development of instruments tailored to the 22q11.2 population could improve assessment precision and allow for more consistent cross-study comparisons.

This review has additional limitations. Variability in participant ages, sample sizes, and assessment instruments complicates synthesis and may introduce bias. Publication bias is possible, as studies with null or negative findings may be underrepresented. Despite these limitations, the review provides a clear and structured overview of the persistent and pervasive nature of language impairments in 22q11.2 syndrome.

Clinically, these findings underscore the importance of early, comprehensive, and individualized assessment and intervention. Programs should address all affected domains—speech, morphosyntax, vocabulary, pragmatics, and literacy—and incorporate multidisciplinary approaches including speech-language therapy, educational support, and psychosocial interventions. Early intervention and therapist training specific to 22q11.2 syndrome are crucial to optimizing outcomes and mitigating the functional impact of these language difficulties. It also underscores the need for greater therapist training regarding the syndrome to enhance the effectiveness of interventions (Moćko et al., 2025). Systematic instruction based on behavioral principles may hold potential for improving communicative and social skills in this population, though further research is needed to guide clinical practice due to the limited number of studies and methodological variability (Roche et al., 2025).

In conclusion, the included studies collectively demonstrate that individuals with 22q11.2 syndrome experience widespread and persistent deficits across speech, language, and communication domains, with both expressive and receptive impairments (Fiksinski et al., 2018; McDonald-McGinn et al., 2015; Norkett et al., 2017). Although the severity and presentation vary across domains and genetic subtypes, the overall pattern indicates that early identification and intervention are essential (Moćko et al., 2025). Methodological limitations highlight the need for longitudinal, standardized, and comparative research to better understand the developmental trajectory of language in this population and to inform evidence-based clinical practice (Rakonjac, 2024).

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Data availability

No data was used for the research described in the article.

References

Antshel, K., Hier, B., Fremont, W., Faraone, S. V., & Kates, W. (2014). Predicting reading comprehension academic achievement in late adolescents with velo-cardio-facial (22q11.2 deletion) syndrome (VCFS): a longitudinal study. *Journal of Intellectual disability research: JIDR, 58*(10), 926–939. https://doi.org/10.1111/jir.12134

Boerma, T., Everaert, E., Vlieger, D., Steggink, M., Selten, I., Houben, M., Vorstman, J., Gerrits, E., & Wijnen, F. (2023). Grammatical skills of Dutch children with 22q11.2 Deletion Syndrome in comparison with children with Developmental Language Disorder: Evidence from spontaneous language and standardized assessment. *Frontiers in Communication, 8*. https://doi.org/10.3389/fcomm.2023.1111584

Cortés-Martín, J., Peñuela, N. L., Sánchez-García, J. C., Montiel-Troya, M., Díaz-Rodríguez, L., & Rodríguez-Blanque, R. (2022). Deletion Syndrome 22q11.2: A Systematic Review. *Children, 9*(8), 1168. https://doi.org/10.3390/children9081168

Everaert, E., Boerma, T., Selten, I., Vorstman, J., & Wijnen, F. (2021). Learning from atypical development: A systematic review of executive functioning in children and adolescents with the 22q11.2 deletion syndrome. *Developmental Review, 60*, Article 100962. https://doi.org/10.1016/j.dr.2021.100962

Everaert, E., Selten, I., Boerma, T., Houben, M., Vorstman, J., de Wilde, H., Derksen, D., Haverkamp, S., Wijnen, F., & Gerrits, E. (2023). The Language Profile of Preschool Children With 22q11.2 Deletion Syndrome and the Relationship With Speech Intelligibility. *American Journal of Speech-Language Pathology, 32*(1), 128–144. https://doi.org/10.1044/2022_AJSLP-21-00328

Fiksinski, A. M., Schneider, M., Murphy, C. M., Armando, M., Vicari, S., Canyelles, J. M., Gothelf, D., Eliez, S., Breetvelt, E. J., Arango, C., & Vorstman, J. A. S. (2018). Understanding the pediatric psychiatric phenotype of 22q11.2 deletion syndrome. *American Journal of Medical Genetics Part A, 176*(10), 2182–2191. https://doi.org/10.1002/ajmg.a.40387

Frascarelli, M., Padovani, G., Buzzanca, A., Accinni, T., Carlone, L., Ghezzi, F., Lattanzi, G. M., Fanella, M., Putotto, C., Di Bonaventura, C., Girardi, N., Pasquini, M., Biondi, M., & Di, F. (2020). Social cognition deficit and genetic vulnerability to schizophrenia in 22q11 deletion syndrome. *Annali dell'Istituto superiore di sanita, 56*(1), 107–113. https://doi.org/10.4415/ANN_20_01_15

Glaser, B., Mumme, D. L., Blasey, C., Morris, M. A., Dahoun, S. P., Antonarakis, S. E., Reiss, A. L., & Eliez, S. (2002). Language skills in children with velocardiofacial syndrome (deletion 22q11.2). *The Journal of Pediatrics, 140*(6), 753–758. https://doi.org/10.1067/mpd.2002.124774

Hamsho, N., Antshel, K. M., Eckert, T. L., & Kates, W. R. (2017). Childhood predictors of written expression in late adolescents with 22q11.2 deletion syndrome: a longitudinal study. *Journal of Intellectual disability research: JIDR, 61*(5), 501–511. https://doi.org/10.1111/jir.12370

Jalal, R., Nair, A., Lin, A., Eckfeld, A., Kushan, L., Zinberg, J., Karlsgodt, K. H., Cannon, T. D., & Bearden, C. E. (2021). Social cognition in 22q11.2 deletion syndrome and idiopathic developmental neuropsychiatric disorders. *Journal of Neurodevelopmental Disorders, 13*(1), 15. https://doi.org/10.1186/s11689-021-09363-4

Kambanaros, M., & Grohmann, K. K. (2017). Linguistic and nonverbal abilities over time in a child case of 22q11 deletion syndrome. *Biolinguistics, 11*, 57–82. https://doi.org/10.5964/bioling.9077

Lorena, J. E., & Sandra, P. R. (2023). Description of Neuropsychological Profile in Patients with 22q11 Syndrome. *Genes, 14*(7), 1347. https://doi.org/10.3390/genes14071347

McDonald-McGinn, D. M., Sullivan, K. E., Marino, B., Philip, N., Swillen, A., Vorstman, J. A., Zackai, E. H., Emanuel, B. S., Vermeesch, J. R., Morrow, B. E., Scambler, P. J., & Bassett, A. S. (2015). 22q11.2 deletion syndrome. *Nature Reviews Disease primers, 1*, Article 15071. https://doi.org/10.1038/nrdp.2015.71

Moraleda-Sepúlveda, E., López-Resa, P., Santos-Muriel, N., López-Peces, P., Frontera, R., & Pulido-García, N. (2023). Implications in Educational, Linguistic, and Psycho-Emotional Development in People with 22q11 Syndrome. *International Journal of Learning and Development, 13*(1), 1. https://doi.org/10.5296/ijld.v13i1.20698

Morrison, S., Chawner, S. J. R. A., Van Amelsvoort, T. A. M. J., Swillen, A., Vingerhoets, C., Vergaelen, E., Linden, D. E. J., Linden, S., Owen, M. J., & Van Den Bree, M. B. M. (2020). Cognitive deficits in childhood, adolescence and adulthood in 22q11.2 deletion syndrome and association with psychopathology. *Translational Psychiatry, 10*(1), 53. https://doi.org/10.1038/s41398-020-0736-7

Moćko, N., Rudzki, M., Miodońska, Z., Olesiak, J., Jochymczyk-Woźniak, K., & Kręcichwost, M. (2025). Development of Speech and Communication in Polish Children with 22q11.2 Deletion Syndrome: A Cross-Sectional Study. *Brain Sciences, 15*(1), 24. https://doi.org/10.3390/brainsci15010024

Norkett, E. M., Lincoln, S. H., Gonzalez-Heydrich, J., & D'Angelo, E. J. (2017). Social cognitive impairment in 22q11 deletion syndrome: A review. *Psychiatry Research, 253*, 99–106. https://doi.org/10.1016/j.psychres.2017.01.103

Persson, C., Niklasson, L., Oskarsdóttir, S., Johansson, S., Jönsson, R., & Söderpalm, E. (2006). Language skills in 5-8-year-old children with 22q11 deletion syndrome. *International Journal of Language & Communication Disorders, 41*(3), 313–333. https://doi.org/10.1080/13682820500361497

Rakonjac, M., Cuturilo, G., Kovacevic-Grujicic, N., Simeunovic, I., Kostic, J., Stevanovic, M., & Drakulic, D. (2024). Speech Sounds Production, Narrative Skills, and Verbal Memory of Children with 22q11.2 Microdeletion. *Children (Basel, Switzerland), 11*(4), 489. https://doi.org/10.3390/children11040489

Rakonjac, M., Cuturilo, G., Stevanovic, M., Jelicic, L., Subotic, M., Jovanovic, I., & Drakulic, D. (2016a). Differences in speech and language abilities between children with 22q11.2 deletion syndrome and children with phenotypic features of 22q11.2 deletion syndrome but without microdeletion. *Research in Developmental Disabilities, 55*, 322–329. https://doi.org/10.1016/j.ridd.2016.05.006

Rakonjac, M., Cuturilo, G., Stevanovic, M., Jovanovic, I., Jelicic-Dobrijevic, L., Mijovic, M., & Drakulic, D. (2016b). Speech and language abilities of children with the familial form of 22q11. 2 deletion syndrome. *Genetika, 48*(1), 57–72. https://doi.org/10.2298/GENSR1601057R

Roche, L., Gale, M., Campbell, L. E., & Sigafoos, J. (2025). Narrative review of communication and social skills interventions for people with 22q11.2 deletion syndrome. *Evidence-Based Communication Assessment and Intervention, 18*(1), 1–12. https://doi.org/10.1080/17489539.2024.2433439

Scherer, N. J., D'Antonio, L. L., & Kalbfleisch, J. H. (1999). Early speech and language development in children with velocardiofacial syndrome. *American Journal of Medical Genetics, 88*(6), 714–723. https://doi-org.bucm.idm.oclc.org/10.1002/(SICI)1096-8628(19991215)88:6<714::AID-AJMG24>3.0.CO;2-B.

Scherer, N. J., D'Antonio, L. L., & Rodgers, J. R. (2001). Profiles of communication disorder in children with velocardiofacial syndrome: Comparison to children with Down syndrome. *Genetics in Medicine, 3*(1), 72–78. https://doi.org/10.1097/00125817-200101000-00016

Schneider, M., Debbané, M., Bassett, A. S., Chow, E. W. C., Fung, W. L. A., Van Den Bree, M. B. M., Owen, M., Murphy, K. C., Niarchou, M., Kates, W. R., Antshel, K. M., Fremont, W., McDonald-McGinn, D. M., Gur, R. E., Zackai, E. H., Vorstman, J., Duijff, S. N., Klaassen, P. W. J., Swillen, A., Gothelf, D., Green, T., Weizman, A., Van Amelsvoort, T., Evers, L., Boot, E., Shashi, V., Hooper, S. R., Bearden, C. E., Jalbrzikowski, M., Armando, M., Vicari, S., Murphy, D. G., Ousley, O., Campbell, L. E., Simon, T. J., Eliez, S., & International Consortium on Brain and Behavior in 22q112 Deletion Syndrome. (2014). Psychiatric Disorders From Childhood to Adulthood in 22q11.2 Deletion Syndrome: Results From the International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome. *American Journal of Psychiatry, 171*(6), 627–639. https://doi.org/10.1176/appi.ajp.2013.13070864

Sebastián Lázaro, D., Brun-Gasca, C., & Fornieles Deu, A. (2020). Lenguaje de niños y jóvenes con síndrome de deleción 22q11. *Revista Délelőtt Neurologia, 71*(03), 99. https://doi.org/10.33588/rn.7103.2020145

Sebastián-Lázaro, D., Brun-Gasca, C., & Fornieles, A. (2022). Comunicación en personas con el síndrome de deleción de 22q11: voz y habla. *Revista Délelőtt Logopedia, Foniatría York Audiología, 42*(4), 238–249. https://doi.org/10.1016/j.rlfa.2021.12.001

Selten, I., Boerma, T., Everaert, E., Vansteensel, M. J., Vorstman, J., & Wijnen, F. (2021). Narrative comprehension and production abilities of children with 22q11.2 deletion syndrome. *Research in Developmental Disabilities, 119*. https://doi.org/10.1016/j.ridd.2021.104109

Solot, C. B., Gerdes, M., Kirschner, R. E., McDonald-McGinn, D. M., Moss, E., Woodin, M., Aleman, D., Zackai, E. H., & Wang, P. P. (2001). Communication issues in 22q11.2 deletion syndrome: children at risk. *Genetics in medicine: official journal of the American College of Medical Genetics, 3*(1), 67–71. https://doi.org/10.1097/00125817-200101000-00015

Solot, C. B., Knightly, C., Handler, S. D., Gerdes, M., Mcdonald-Mcginn, D. M., Moss, E., Wang, P., Cohen, M., Randall, P., Larossa, D., Driscoll, D., Emanuel, B., & Zackai, E. H. (2000). Communication disorders in the 22Q11. 2 microdeletion syndrome. *Journal of Communication Disorders, 33*(3), 187–204. https://doi.org/10.1016/S0021-9924(00)00018-6

Solot, C. B., Sell, D., Mayne, A., Baylis, A. L., Persson, C., Jackson, O., & McDonald-McGinn, D. M. (2019). Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management. *American Journal of Speech-Language Pathology, 28*(3), 984–999. https://doi.org/10.1044/2019_AJSLP-16-0147

Van Den Heuvel, E., Manders, E., Swillen, A., & Zink, I. (2017a). Parental report on socio-communicative behaviours in children with 22q11.2 deletion syndrome. *Journal of Intellectual & Developmental Disability, 42*(2), 162–172. https://doi.org/10.3109/13668250.2016.1213796

Van Den Heuvel, E., Manders, E., Swillen, A., & Zink, I. (2018). Atypical language characteristics and trajectories in children with 22q11.2 deletion syndrome. *Journal of Communication Disorders, 75*, 37–56. https://doi.org/10.1016/j.jcomdis.2018.06.001

Van Den Heuvel, E., ReuterskiöLd, C., Solot, C., Manders, E., Swillen, A., & Zink, I. (2017b). Referential communication abilities in children with 22q11.2 deletion syndrome. *International Journal of Speech-Language Pathology, 19*(5), 490–502. https://doi.org/10.1080/17549507.2016.1221456

Verbesselt, J., Solot, C. B., Van Den Heuvel, E., Crowley, T. B., Giunta, V., Breckpot, J., McDonald-McGinn, D. M., Zink, I., & Swillen, A. (2023). Language Profiles of School-Aged Children with 22q11.2 Copy Number Variants. *Genes, 14*(3), 679. https://doi.org/10.3390/genes14030679

Verbesselt, J., Van Den Heuvel, E., Breckpot, J., Zink, I., & Swillen, A. (2022). Parent-Reported Social-Communicative Skills of Children with 22q11.2 Copy Number Variants and Siblings. *Genes, 13*(10), 1801. https://doi.org/10.3390/genes13101801

Wierzchowski, A., Sablich-Duley, S., & Bordes Edgar, V. (2021). Variability in Neuropsychological Phenotypes in Patients with 22Q11.2 Deletion Syndrome: Case Series. *Developmental Neuropsychology, 46*(5), 381–392. https://doi.org/10.1080/87565641.2021.1956498

Zinkstok, J. R., Boot, E., Bassett, A. S., Hiroi, N., Butcher, N. J., Vingerhoets, C., Vorstman, J. A. S., & Van Amelsvoort, T. A. M. J. (2019). Neurobiological perspective of 22q11.2 deletion syndrome. *The Lancet Psychiatry, 6*(11), 951–960. https://doi.org/10.1016/S2215-0366(19)30076-8