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Analysis of the pragmatic competence profile in the population with 22q11.2 syndrome: a comparison between syndromic presentations

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Introduction: Various studies confirm that individuals with 22q11.2 syndrome exhibit communicative alterations that affect their social interactions. However, few compare the pragmatic characteristics of these individuals based on the type of syndrome (either deletion or duplication).
Method: This research aims to address the pragmatic skills of 10 participants with 22q11.2 syndrome, comparing the types 22q11.2DupS and 22q11.2DS, within an age range of 7 to 17 years, thereby confirming or denying communicative differences between syndromic presentations. The evaluation was conducted using the Revised Quick Pragmatic Assessment Protocol, allowing observation of the communicative characteristics of participants at the textual, utterance, and interactive levels. Video recordings of conversational samples with close interlocutors were used for the evaluation.
Results: The results show that, in general, there are differences between the two types of the syndrome, with more pragmatic difficulties in the case of people 22q11.2DS.
Conclusion: These differences do not appear to be related to age, as the percentage of different items is similar across both age ranges. Some limitations of the study are discussed.

KEYWORDS

22q11.2, pragmatics, deletion, duplication, PREP-R

Introduction

22q11.2 syndrome, also known as velocardiofacial syndrome or DiGeorge syndrome (Conley et al., 1979; DiGeorge, 1965), is a genetic condition characterized by the loss (22q11.2DS) or duplication (22q11.2DupS) of genetic material in the q11.2 region of chromosome 22 (Bartik et al., 2022; Driscoll, 2001). While 22q11.2DS is the more prevalent form, with an estimated incidence of between 1 in 2,000 and 1 in 4,000 live births (Blagojevic et al., 2021; Devriendt et al., 1998; Driscoll, 2001), there is some uncertainty regarding the actual prevalence of 22q11.2DupS, which is considerably less studied due to lower medical detection rates and fewer apparent congenital anomalies in these individuals (Olsen et al., 2018). Both syndromes are characterized by the presence of congenital heart defects, dysmorphic facial features, cleft palate, and linguistic, educational, and psychological-psychiatric difficulties that significantly reduce quality of life (Álvarez et al., 2009; Cortés-Martín et al., 2022; Driscoll, 2001; Yu et al., 2019). Additionally, cognitively,

individuals with 22q11.2 exhibit intellectual capabilities in the borderline range, with an IQ between 70 and 85, and there may be cases where IQ falls between 55 and 70 (Drmic et al., 2022; Gerdes et al., 1999).

Studies on 22q11.2 syndrome suggest difficulties in the lexical-semantic and morphosyntactic components in both populations (Solot et al., 2000; Verbesselt et al., 2023). However, in the population with 22q11.2DS, there are more problems in designating and defining concepts, affecting lexical-semantic skills, while individuals with 22q11.2DupS exhibit morphosyntactic alterations, characterized by short and less complex sentences (Verbesselt et al., 2023).

Lastly, regarding pragmatics, the linguistic alterations present in this population tend to transform into pragmatic difficulties that negatively impact social interactions (Álvarez et al., 2009). Consequently, it has been observed that school-aged children with 22q11.2 syndrome show an inability to use contextual information for understanding, organizing, and expressing language appropriately, often focusing on secondary issues or details instead (Van Den Heuvel et al., 2018).

Non-verbal communication is also affected, as children with 22q11.2 tend to engage in editing tasks in close environments (Sebastián-Lázaro et al., 2020) and do not accurately interpret the facial expressions of their interlocutors to understand meaning in context (Murphy, 2004; Sebastián-Lázaro et al., 2020). Similarly, alterations in prosodic nuances are noted, resulting in reduced and disharmonious speech rhythm (Sebastián-Lázaro et al., 2022; Solot et al., 2000; Van Den Heuvel et al., 2017a).

These contextual difficulties, along with severely impaired intelligibility, result in lower communicative intent, expressed through less frequent communication and fewer statements (Van Den Heuvel et al., 2017a). In conclusion, it can be inferred that the enunciative pragmatics are altered due to a violation of the maxims proposed by Grice (1975).

Individuals with 22q11.2 deletion syndrome exhibit significant pragmatic impairments that affect their communicative competence in social contexts. These difficulties include challenges in interpreting tone of voice, facial expressions, abstract language, and emotional cues from interlocutors, as well as maintaining discourse coherence and conversational turn-taking. The literature suggests that these pragmatic disorders are secondary to an altered linguistic profile rather than primary social deficits, as difficulties are observed in semantic fluency, discourse organization, and lexical retrieval (Sebastián-Lázaro et al., 2020; De Smedt et al., 2007). The range of observed disorders includes problems with non-verbal communication, irrelevant or out-of-context verbal interventions, limited use of discourse connectors, and unnecessary visual details. Research primarily focuses on expressive and receptive language, semantic fluency, and emotional comprehension, assessed through standardized psychometric tests and parent questionnaires, although complementary methods such as clinical observation and spontaneous discourse analysis are increasingly recommended (Van Den Heuvel et al., 2017b). Participants are typically children and adolescents aged 5 to 21 years, often with borderline intellectual functioning or mild intellectual disability, which influences their pragmatic performance.

Regarding interactive pragmatics, individuals with 22q11.2 often have difficulties respecting turn-taking due to anxiety and time pressure, leading to multiple overlaps and interruptions during interactions (Sebastián-Lázaro et al., 2020). Theoretically, pragmatics refers to the use of language according to context and speaker intention, while interactive pragmatics emphasizes the dynamic aspects of interaction, such as meaning negotiation, conversational cooperation, and adaptation to the interlocutor. This distinction is supported by Grice's cooperative principles, and Sperber and Wilson's relevance theory (Escandell, 2006). As a result, individuals with 22q11.2 seem to struggle with positioning themselves within the adjacent pair during the communicative process (Solot et al., 2019). Additionally, concerning textual pragmatics, they experience challenges in recalling plots, resulting in narratives with incoherent content structure, thematic leakage, and few cohesive elements (Persson et al., 2006; Van Den Heuvel et al., 2017b).

Although scientific literature has highlighted the linguistic difficulties experienced by people with 22q11.2 syndrome in recent years, little research has been done on language differences based on the type of syndromic presentation in this population. In these investigations, Verbesselt et al. (2023) showed that children with 22q11.2DS displayed linguistic difficulties that began at the word level; the most common linguistic problems in children with 22q11.2DS began at the sentence level. Notably, both expressive and receptive language, as well as lexical-semantic and morpho-syntactical domains, were affected in both types of syndromic presentations. However, no studies have been found that specifically focus on the development of the pragmatic component based on the existence of duplication or deletion.

Therefore, the aim of this research is to analyze the communicative profile of individuals with 22q11.2 in a natural context, in order to determine whether there are differences between the presentations of 22q11.2DupS and 22q11.2DS.

Method

Design

A cross-sectional study with a quasi-experimental design was conducted to compare pragmatic language performance between participants with different types of 22q11.2 deletion.

This non-randomized comparison of pre-existing groups was complemented by descriptive and observational strategies to address the research questions. In this way, we observe a dependent variable, a continuous quantitative type corresponding to the level of pragmatic skill, which has three levels: general pragmatic skill, specific pragmatic skill, and grammatical base pragmatic skill. Additionally, two independent variables are observed. First, there is a dichotomous nominal variable corresponding to the type of 22q11.2 syndrome, divided into two levels: deletion and duplication. Second, a second variable is noted, which is of an ordinal quantitative type, corresponding to age, with two levels: between 7 and 12 years, and between 12 and 17 years. Finally, it is worth noting that the various items of the evaluation tool (see Instruments section) were also analyzed individually, which

TABLE 1 Participants divided by groups and mean age.

Groups	22q11.2DupS (7–12 years)	22q11.2DS (7–12 years)	22q11.2DupS (12–17 years)	22q11.2DS (12–17 years)
Number of participants	2	2	3	3
Mean age	9.09	10.1	15.9	15.5
Standard deviation (SD)	2.64	2.68	1.73	2.3

constitute nominal qualitative variables with three response levels: yes, no, and not assessable.

Participants

The initial sample consisted of 12 participants. However, 2 of them were excluded because they did not meet the established inclusion criteria for the study, which will be explained later. Thus, the final sample for this study comprised 10 participants (5 males and 5 females), aged between 7 and 17 years, with a mean age of 13.3 (SD = 3.69). This group was divided based on the syndromic presentation.

Recruitment was conducted through convenience sampling in collaboration with 22q11.1 Spanish Association, which limited the pool of eligible participants. The sex ratio in our sample (1:1) does not necessarily reflect the exact distribution in the general population with 22q11.2DS, but rather the composition of the accessible sample during the recruitment period.

The first group consisted of 5 individuals with 22q11.2DupS (3 females and 2 males), with a mean age of 13.2 (SD = 4.17), while the second group comprised 5 individuals with 22q11.2DS (2 females and 3 males), with a mean age of 13.7 (SD = 4.12). Additionally, each group was further subdivided into 2 age ranges (7 to 12 years and 12 to 17 years), resulting in 4 participants (2 from each group) in the 7–12 age range, with a mean age of 9.59 (SD = 2.25), and 6 participants (3 from each group) in the 12–17 age range, with a mean age of 15.7 (SD = 1.84). This information is presented visually in the following table (Table 1).

To obtain a comparable analysis of results, a comparison was made between each type (participants with 22q11.2DS and users with 22q11.2DupS), further divided into the age range of 7 to 12 years and the age range of 12 to 17 years. Given the broad age range, there may be differences at the cognitive, linguistic, and emotional levels, with participants in the first group having a simpler language and social environment than those in the second group, who communicate in a wider and more complex context. For these reasons, it was decided to divide the participants into two age ranges to try to control this extraneous variable.

To collect the sample, the inclusion criteria established were that participants had a diagnosis of 22q11.2 made by a medical specialist, had a minimum verbal competence to be evaluated, were Spanish speakers, that the videos displayed the characteristics explained when contacting the participants (detailed further in the Procedure section), absence of comorbidities, and that both their legal guardians and the participants themselves provided consent to conduct the research. Exclusion criteria included participants who did not have a clear syndromic specificity in the diagnosis of 22q11.2 Syndrome or the presence of other disorders and comorbidities. It is important to note that all participants had an Intelligence Quotient (IQ) in the range of 70–79, ensuring that they were equated based on IQ (p s> 0.05).

Instruments

The Revised Quick Pragmatic Assessment Protocol (PREP-R) (Gallardo Paúls et al., 2015) was used, an instrument composed of items divided and explained according to the classification of pragmatic types proposed by Gallardo-Paúls (2009) and discussed in the theoretical framework (enunciative pragmatics, textual pragmatics, and interactive pragmatics). It also allows for differentiation between communicative problems arising from deficits in language components (grammatical base pragmatic deficits) and communicative problems resulting from specific pragmatic deficits. Overall, the global assessment refers to the general pragmatic skill of each evaluated subject but allows for the calculation of percentages of preserved specific pragmatic skill and grammatical base pragmatic skill.

This qualitative test enables the analysis and evaluation of the skills and/or difficulties an individual may present in interactions within their everyday ecological environment, recording difficulties and the strategies or behaviors the speaker uses to compensate for or mask these communicative limitations. It is divided into 18 items organized into three levels of pragmatic analysis: 6 items for enunciative pragmatic evaluation, 5 items for textual pragmatic evaluation and 7 items for interactive pragmatic evaluation.

The enunciative and textual levels are grouped into sublevels within each level. In this regard, the enunciative level has three sublevels (Speech Acts, Editing Tasks, and Inferences), and the textual level has two (Coherence and Cohesion), while the interactive level is not divided into sublevels. Furthermore, when a single item requires examination of several aspects, these are organized into sub-items, and each item and sub-item includes a brief explanation to guide the evaluator and remind them of the behaviors to observe in each case.

The evaluation is conducted through systematic analysis of language samples, typically collected in naturalistic or semi-structured contexts. Each item is scored based on observed communicative behaviors, and the results are expressed as percentages of preserved ability in three domains: general pragmatic ability, specific pragmatic ability, and grammar-based pragmatic ability. These percentages, as shown in Tables 2, 3, are calculated by dividing the number of items scored positively in each domain by the total number of items assessed, providing a profile of strengths and weaknesses.

TABLE 2 Percentages of pragmatic skills in the age range of 7 to 12 years.

Group	General pragmatic skill (GPS)	Average percentage of GPS	Specific pragmatic Skill (SPS)	Average percentage of SPS	Grammatical base pragmatic skill (GBPS)	Average percentage of GBPS
22q11.2DupS	67%	68%	59%	66%	86%	80%
	69%		74%		75%	
22q11.2DS	88%	85%	88%	84%	87%	87%
	81%		80%		86%	

TABLE 3 Percentages of pragmatic skills in the age range of 12 to 17 years.

Group	General pragmatic skill (GPS)	Average percentage of GPS	Specific pragmatic skill (SPS)	Average percentage of SPS	Grammatical base pragmatic skill (GBPS)	Average percentage of GBPS
22q11.2DupS	65%	75%	50%	70%	100%	92%
	87%		82%		100%	
	72%		79%		75%	
22q11.2DS	71%	60%	76%	60%	57%	61%
	85%		79%		100%	
	24%		26%		25%	

Given its central role in ensuring the accuracy and reliability of the data, transcription was undertaken in accordance with the main conventions of the PerLa Corpus (Fernández-Urquiza and Gallardo-Paúls, 2015). These conventions comprise the verbatim reproduction of utterances, the use of square brackets to indicate overlapping speech, ellipses for pauses or unfinished utterances, capitalization to mark prosodic emphasis, and standardized symbols for non-verbal elements. Adhering to these criteria reinforced the methodological rigor of the study and facilitated the interpretation of the illustrative examples.

The PREP-R has been applied primarily in clinical populations with language impairments, including children with neurodevelopmental disorders. In our study, it was used to explore pragmatic profiles in individuals with 22q11.2 deletion and duplication syndromes. We will revise the methodology section to include a clearer description of the scoring system, the rationale for using this tool, and its application context. The use of this test has been studied to assess pragmatics in other populations with intellectual disabilities, such as Down syndrome (Moreno and Díaz, 2014) and Williams syndrome (Shiro et al., 2016).

Procedure

First, a review of the existing scientific literature was conducted to establish the theoretical framework for the research, corresponding with the introduction part. This analysis confirmed the need to study pragmatics in individuals with 22q11.2 syndrome, specifically in comparison between the two existing types. Following the information search, the documentation was sent to the ethics committee, and upon approval, data collection for the participants began along with the distribution of informed consent.

Subsequently, a text message was drafted for the families of the subjects, detailing the procedures to be followed and explaining the nature of their participation. The message was disseminated via email to potential participants who met the selection criteria, and whose information was provided by principal investigator. It is important to note that prior to the data transfer, consent was signed regarding data confidentiality and the use of the research data.

Video recordings lasting between 10 and 15 min were requested, in which the individual with 22q11.2 should be seen interacting with a family member or someone with whom they spent the most time. The decision to use video recording was made as it is the most recommended method in the protocol guidelines (Bertrán et al., 2018). The use of video recordings to capture spontaneous language samples was essential for ensuring the accuracy, richness, and reliability of the pragmatic analysis. Unlike live observation or audio-only formats, video allowed for the detailed review of both verbal and non-verbal communicative behaviors, such as facial expressions, gestures, eye contact, and turn-taking dynamics. This multimodal perspective is particularly important when evaluating pragmatic competence, as it provides contextual cues that are crucial for interpreting the speaker's intentions and interactional strategies. Additionally, video recordings enabled repeated viewing and collaborative coding among researchers, which strengthened the consistency of the evaluations and facilitated a more nuanced understanding of the participants' communicative profiles.

Furthermore, the recordings took place at home, as this was where the participants felt comfortable conducting conversational activities. Specifically, individuals were to engage in conversations that were as natural as possible with an interlocutor. Once the families completed the videos, they sent the corresponding material to the study's principal investigator via an encrypted link. The

recordings were taken into account in their entirety in an attempt to establish a more comprehensive and complete pragmatic profile.

Previously, socio-family and educational data were also collected from the families. All families belonged to a middle-income bracket, and the parents' educational attainment was middle-to-high.

Ethical aspects

This study has been approved by the Social Research Ethics Committee of UCLM under reference CAU-683200-X6H7. Thus, the study adheres to the ethical principles outlined in the Declaration of Helsinki regarding research with human beings, as well as to current Spanish legislation which stipulates that participants must have the necessary information about the project to decide whether or not they wish to participate. In this regard, all subjects were thoroughly and properly informed about the process, ensuring that their participation in the study was voluntary. However, since all participants were minors, authorization from their parents or legal guardians was required, who were also properly informed.

Regarding confidentiality and data protection, informed consent was developed and signed, taking into account the confidentiality guarantees established by the Spanish's laws. Only the research team had access to the collected and pseudonymized data, which were maintained anonymously, meaning that no names or identifying details were included other than the age of each participant. In this way, the confidentiality of all study participants is fully guaranteed.

Data analysis

Ten video recordings were analyzed, one for each case. The total analysis period was 8 weeks, with the first 2 weeks dedicated to data transcription following the conventions established by the PerLa corpus (Gallardo Paúls and Veyrat Rigat, 2004), which provided uniformity in the transcribed representation of the data for subsequent evaluation. In the following 5 weeks, each video underwent individual analysis using the PREP-R protocol by each evaluator, assessing both each item proposed in the protocol and the percentages of pragmatic skills. Finally, in the last week of analysis, an inter-rater agreement process was conducted.

Due to the qualitative and subjective nature of the PREP-R, an inter-rater evaluation was conducted with three members. This methodology involves comparing and averaging the results of one evaluator with those of another. This approach helps to avoid biases when comparing the assessment of each criterion by three different evaluators. The Kappa coefficient of Cohen was used to analyze the degree of agreement among the evaluations, given that the variables were nominal. Following a thorough individual analysis of each conversational sample, a 69% agreement was obtained. After this, a meeting was held with the three evaluators to discuss the disagreements, resulting in a final agreement of 97% in the outcomes. Lastly, to establish a comparison between the groups, and as shown subsequently in Tables 1, 2, a weighted average of the percentages of pragmatic skills described in the PREP-R was calculated, visually displaying the differences and indicating which aspects each group scored higher in comparison to their counterparts with the other type of syndrome.

Results

The results have revealed that in both general and specific pragmatic skills, the group of individuals with 22q11.2DS scored higher than the group with 22q11.2DupS in the age range of 7 to 12 years, while the percentages of grammatical base pragmatic skills were very similar (Table 2).

In the age range of 12 to 17 years, a superiority is observed in the percentages of all evaluated pragmatic skills among individuals with 22q11.2DupS compared to participants with 22q11.2DS (Table 3).

The results obtained are a product of the analysis using the PREP-R. However, it is important to note that there were items that could not be evaluated in all situations due to the characteristics of the interaction not allowing for the specific aspect to be assessed. The following discusses the different items based on the type of pragmatics they evaluate.

Firstly, the results indicate an uneven profile between individuals with 22q11.2DupS and 22q11.2DS in the components of enunciative pragmatics. In this regard, it can be observed that the group with 22q11.2DupS in the age range of 7 to 12 years shows a higher percentage in the production of enunciative acts. In contrast, there are no differences between both groups in the comprehension and/or production of propositional acts, pauses and intraturn silences, direct speech acts, indirect speech acts, locutive acts, or draft acts, as shown in Extract 1.

The following are several examples between the reference adult (A) and the person with 22q11.2 (Q).

Extract 1

Example of the use of a verbal strategy that allows for gaining time for the construction of utterances (draft act).

User 12, 8 years old, with 22q11DS

- A: What about the noodles?
- Q: These ramen ones that come in a—one—uh, this thing I don't know what it's called, a container.
- A: What are those noodles like?
- Q: They're noodles that are like this long.

In the age range of 12 to 17 years, the results show that the group consisting of individuals with 22q11.2DupS also achieves a higher percentage in the production of enunciative acts and in the use of pauses and intraturn silences. Additionally, unlike the previous age range, in this range we find that the group with 22q11.2DS has a higher percentage in the comprehension and/or production of direct speech acts compared to the group with 22q11.2DupS. In the age range of 12 to 17 years, as in the previous age interval, no differences are found in the comprehension and/or production of propositional acts, indirect speech acts, as shown in Extract 2, locutive acts, and draft acts.

Extract 2

Example of correct production of an indirect speech act.
User 2, 17 years old, with 22q11DS

- *A: The Power Rangers, and the shows you used to watch on those videos you had, remember?*
- *Q: Yes, on Boing—they always played them in the summer because every time I watched Boing in the summer, I would always tell my brother, "COME ON ALONSO, LET'S PUT ON BOING because they're showing the Power Rangers now, I don't know what..." Well, every time we traveled—every time we were coming back from a trip—we would always either stay up late traveling or we would always take out the tablet and watch YouTube or whatever.*

Continuing with the paralinguistic elements and editing tasks, the results do not indicate any differences between individuals with 22q11.2DupS and 22q11.2DS in the age range of 7 to 12 years, except in the use of compensatory gesturing, where the group with 22q11.2DupS achieved a higher percentage. In contrast, it was the group with 22q11.2DS that obtained the highest percentage in rectification capacity and metapragmatic awareness (Extract 3).

Extract 3

Example of rectification in the production of a statement.
User 4, 11 years old, with 22q11DS
Q: Eeh/ I'm 9 years old, no - I'm 10 years old - 11 years and 9 months and ///

On the other hand, in the age range of 12 to 17 years, the use of compensatory gesturing is the only item in the editing tasks where differences are found, with the group with 22q11.2DS standing out compared to the group with 22q11.2DupS (Extract 4).

Extract 4

Example of a gesture that regulates verbal production (compass regulator).
User 2, 17 years old, with 22q11DS

- *A: Yesterday was the presentation and today you start it.*
- *Q: And every week we cover a topic (makes a gesture with one finger) from that course.*
- *A: And you have to do...*
- *Q: An exam and some activities on that topic.*

To conclude, regarding the principle of conversational cooperation, the results indicate that in the age range of 7 to 12 years, the group with 22q11.2DS commits fewer violations of the maxims of quality, manner, and relevance, as well as of particularized implicatures (Extract 5). No differences were observed in the maxim of quantity or in conventional implicit acts, also known as lexicalized expressions or idioms.

Extract 5

Example of a violation of the maxim of manner through ambiguous verbal production.
User 12, 8 years old, with 22q11DS

- *A: The Great Wall of China! Uh, how do you play that? I really don't know anything at all.*
- *Q: Look, one person has to stand in the middle, and everyone else—the one in the center has to say "wall," and the others have to say "China," and we have to start running so they don't catch us // (gestures exhaustion).*
- *A: And what?*
- *Q: And you have to get to the other side. You have to say "Great Wall of China" every time and then run away, and that's it.*

However, in the age range of 12 to 17 years, we find a less homogeneous profile. While the group with 22q11.2DupS commits fewer violations of the maxims of quality, manner, and relation, they produce more violations of the maxim of quantity and particularized implicatures compared to the group with 22q11.2DS (Extract 6). On the other hand, no differences were found in lexicalized expressions or idioms.

Extract 6

Example of a violation of the maxim of quantity due to insufficiency.
User 7, 14 years old, with 22q11DupS

- *A: Okay, how's school going // in high school?*
- *Q: Good.*
- *A: Do you have many friends?*
- *Q: Yes.*
- *A: And girl friends?*
- *Q: No.*

In Table 4, the enunciative differences are presented visually, showing the percentages of each participant group for each item.

Continuing with textual pragmatics, in the age range of 7 to 12 years, specifically within the coherence sublevel, the group of individuals with 22q11.2DS demonstrates a higher percentage in the narrative superstructure item (Extract 7). On the other hand, there are no differences between the two groups regarding argumentative superstructure, recognition of a new topic, or the introduction of a thematic shift.

Extract 7

Example of Incorrect Use of Narrative Superstructure Due to Inadequate Character Presentation.
User 1, 12 years old with 22q11DS

- *Q: No, it's Raquel; she loves you more, Barbie.*
- *A: Who is Raquel? I don't know who Raquel is.*

TABLE 4 Characteristics of enunciative pragmatics according to PREP-R items.

Ítems	Group 22q11.2DupS (7–12 years)	Group 22q11.2DS (7–12 years)	Group 22q11.2DupS (12–17 years)	Group 22q11.2DS (12–17 years)
Enunciative acts	100%	0%	100%	66.7%
Propositional acts	100%	100%	100%	100%
Intra-turn pauses and silences	100%	100%	100%	66.7%
Direct speech acts	50%	50%	33%	67%
Indirect speech acts	50%	50%	67%	67%
Locutionary acts	100%	100%	100%	100%
Erasing acts	100%	100%	100%	100%
Compensatory gestures	100%	50%	67%	100%
Rectification and metapragmatic awareness	50%	100%	67%	67%
Quality implicature	50%	100%	67%	33%
Quantitative implicature	0%	0%	0%	33%
Manner implicature	0%	100%	67%	33%
Relation implicature	50%	100%	67%	33%
Specific implicatures	50%	100%	33%	67%
Lexicalized expressions or idioms	50%	50%	67%	67%

- *Q: A friend of Barbie's who has a brother named Ryan, who is Ken's friend.*
- *A: Oh, okay, okay, okay, do we have that Raquel with us?*
- *Q: No.*

Unlike the previous age interval, in the group aged 12 to 17 years, individuals with 22q11.2DupS show a higher percentage compared to their peers with 22q11.2DS in items related to narrative superstructure, recognition of a new theme or thematization, and appropriate thematic change. In contrast, the results do not show differences between the two groups in the item related to argumentative superstructure.

Regarding the sublevel of cohesion, in the age group of 7 to 12 years, there is a higher percentage of lexical effectiveness in the 22q11.2DS group compared to individuals with 22q11.2DupS (Extract 8). However, with respect to morphology, word formation, syntax, and grammatical construction, no differences are found between the two groups.

Extract 8

Example of correct lexical effectiveness, without repetitions or empty words.

User 4, 11 years old with 22q11DS

- *A: What's it about? Because Minecraft is a big topic to discuss.*
- *Q: (LAUGHS) Uh, about Minecraft—well, Minecraft is mainly for building, for imagination, and of course, the game is… is it normal? No, it's all made of squares. I mean, yes, although some things can be round, but no—everything has square pixels. So there's nothing round, or rectangular, or anything.*

On the other hand, the group of individuals with 22q11.2DupS in the age range of 12 to 17 years shows a higher percentage in all items related to textual cohesion, that is, in lexical effectiveness, morphology and word formation, as well as syntax and grammatical construction. Table 5 shows the percentages of groups of individuals with 22q11.2DupS and 22q11.2DS in both age ranges for each of the items related to textual pragmatics.

Lastly, regarding interactive pragmatics, in the age range of 7 to 12 years, we find differences in turn-taking fluency, conversational participation, predictability, and the communicative use of eye contact, where the group of individuals with 22q11.2DS shows a higher percentage. In contrast, in the item related to natural gesturing to complement language (as shown in Extract 9), individuals with 22q11.2DupS scored better compared to participants with 22q11.2DS.

Extract 9

Example of correct use of natural gesturing.

User 6, 7 years old with 22q11DupS

- *Q: I can escape from Manuel.*
- *A: Really?*
- *Q: Yes, I'm running away (gesture of running)*

On the other hand, the results do not show differences between the two types of syndrome concerning the agility and speed of turn-taking and the design of turns based on conversational priority.

Unlike the previous age range, in the interval of 12 to 17 years, there is a higher percentage in the group of individuals with 22q11.2DupS regarding items related to turn agility, turn-taking,

TABLE 5 Characteristics of textual pragmatics according to the items of the PREP-R.

Items	Group 22q11.2DupS (7–12 years)	Group 22q11.2DS (7–12 years)	Group 22q11.2DupS (12–17 years)	Group 22q11.2DS (12–17 years)
Narrative superstructure	0%	50%	33%	0%
Argumentative superstructure	50%	50%	67%	67%
Thematization	100%	100%	100%	100%
Thematic change	50%	50%	67%	33%
Lexical effectiveness	50%	100%	67%	33%
Morphology and word formation	100%	100%	100%	67%
Syntax and grammatical construction	100%	100%	100%	67%

TABLE 6 Characteristics of interactive pragmatics according to the items of the PREP-R.

Items	Group 22q11.2DupS (7–12 years)	Group 22q11.2DS (7–12 years)	Group 22q11.2DupS (12–17 years)	Group 22q11.2DS (12–17 years)
Agility of turn	100%	100%	100%	67%
Turn-taking	50%	100%	67%	33%
Conversational participation	0%	50%	33%	33%
Predictability	50%	100%	33%	0%
Priority	50%	50%	33%	67%
Natural gesturing	100%	50%	67%	67%
Communicative use of eye contact	50%	100%	67%	67%

and predictability. In contrast, individuals with 22q11.2DS design more turns according to the principles of conversational priority than their peers with 22q11.2DupS. However, the results do not show differences between the two types in this age range regarding the conversational participation index, the use of natural gesturing, and the communicative use of eye contact.

Finally, Table 6 shows the percentages for each item related to interactive pragmatics.

Discussion

The study aims to provide novel insights into the pragmatic profile of individuals with 22q11.2 syndrome, establishing differences between the two typologies: 22q11.2DupS and 22q11.2DS. The study highlights that children with 22q11.2DS not only present articulation difficulties, consistent with previous findings on speech sound disorders in this population (Everaert et al., 2023; Persson et al., 2003; Solot et al., 2019), but also exhibit pragmatic impairments that affect their ability to use language effectively in social contexts. These challenges are likely influenced by underlying facial and palatal anomalies (Goldmuntz, 2020; Solot et al., 2001; Yu et al., 2019), which contribute to both structural speech deficits and limitations in expressive communication, further impacting pragmatic functioning. Furthermore, while research by Van Den Heuvel et al. (2017b) indicates that individuals with 22q11.2SD require training to avoid inappropriate pauses in conversations, the results of this study show that most participants manage pauses and intra-turn silences appropriately.

On the other hand, the results align with research indicating that individuals with 22q11.2 syndrome often struggle with tasks involving "theory of mind," which could explain the disparate scores in tasks such as indirect speech acts or the use of lexicalized expressions and idioms (Laorden et al., 2019; Niklasson et al., 2002, 2009). In line with this, Van Den Heuvel et al. (2017a) suggest that individuals with 22q11.2 have difficulty incorporating such speech acts appropriately within context.

Persson et al. (2006) proposed that individuals with 22q11.2 employ editing strategies, such as gestures and paralinguistic cues, due to delays in oral language acquisition. This theory could explain the findings of this study, as most participants consistently and correctly used compensatory behaviors, metapragmatic awareness, and natural gesturing. These results also relate to studies highlighting expressive language poverty compared to receptive language across all ages in individuals with 22q11.2 (Roche et al., 2020; Solot et al., 2019), thus transgressing the maxim of quantity and compensating for difficulties with gestures.

Regarding textual pragmatics, the study shows that a low percentage of individuals can produce coherent narratives, which is consistent with studies indicating that individuals with 22q11.2 exhibit similarities to those with language disorders concerning narrative difficulties (Selten et al., 2021). For instance, Boerma et al. (2023) reported weak narrative skills in children with 22q11.2, particularly regarding macrostructure, which could

explain their challenges in presenting events, characters, and, generally, information in a logical and coherent order.

Regarding thematic management, the results indicate differences between the two types of 22q11.2 syndrome in the age range of 12 to 17 years, with individuals with 22q11.2DS facing more difficulties. These findings do not support the hypothesis of Wenger et al. (2016), who assert that individuals with 22q11.2DupS exhibit restricted interests, which could lead users to repeatedly return to the same topic of conversation (Spiker et al., 2012). In terms of cohesion, the evidence from this research shows that most participants use words and word constructions appropriately, although some specific deficits were noted. This may be attributed to a good command of vocabulary and grammar among participants (Louwerse, 2004), contradicting previous studies that reported difficulties in syntax and discourse organization (Solot et al., 2000; Verbesselt et al., 2023).

The study's results reveal that the majority of participants do not demonstrate adequate conversational engagement, which may be due to a passive and withdrawn conversational style, particularly highlighting the lack of conversational participation by individuals with 22q11.2DS in the 7 to 12 age range (Van Den Heuvel et al., 2017b). Several studies also report challenges in initiating conversations and taking turns (Angkustsiri et al., 2014; Van Den Heuvel et al., 2017b). While the results vary concerning age and type, individuals with 22q11.2DS appear to experience increasing difficulties over time with rising social demands (Sebastián-Lázaro et al., 2020), whereas individuals with 22q11.2DupS may improve their skills as they grow, presenting milder symptoms (Verbesselt et al., 2022). Communicative participation may also be influenced by personal and environmental factors, as well as the interest in the interlocutor (Blum-Kulka et al., 2010; Shea, 2022). Thus, the study's results, in alignment with various studies, suggest that children tend to interact more extensively and confidently with their peers compared to adults, facilitating a reciprocal exchange of idea. Finally, difficulties in the communicative use of gaze were observed in participants with 22q11.2DupS, which could be related to an alteration in the non-linguistic elements of communication, similar to individuals with ASD (Fernández and García, 2020; Wenger et al., 2016).

In summary, the study has allowed us to conclude that there are pragmatic differences between individuals with 22q11.2DupS and 22q11.2DS. However, certain limitations of the study necessitate further investigation in this area, such as the limited sample size due to the low prevalence of the syndrome and the geographic distribution of participants. In relation to this limitation, it would be valuable to evaluate a broader sample concerning age and social context, taking into account differences in family or educational environments.

In conclusion, the results reveal differences between individuals with 22q11.2DupS and 22q11.2DS across most items in the PREP-R protocol. However, these differences do not align with the theory that individuals with 22q11.2DupS present fewer difficulties due to less severe symptoms than those with 22q11.2DS, as the findings do not allow us to reach a consensus regarding the pragmatic severity of one profile compared to the other. Therefore, we can conclude that the main hypothesis posited is fulfilled, establishing differences between both subtypes in the 7 to 12-year age range as well as in the 12 to 17-year interval, with no significant variations between these two age ranges. These results underscore the need for speech therapy intervention within this population to enhance communicative proficiency and, consequently, to improve social skills and the quality of life for both individuals with 22q11.2 syndrome and their regular interlocutors. Pragmatic research on the syndrome is limited, highlighting an area of knowledge that requires further study to facilitate early diagnosis and the initiation of effective treatments, thereby improving the quality of life of individuals and their communities, opening up potential new avenues for research.

Limitations of the study

As possible limitations of the present study, it should be noted that the sample size could have been larger and that the results are primarily based on Spanish speakers, which does not allow us to generalize these findings to other languages. In addition, the duration of the video recordings may not have been sufficient to capture the full range of pragmatic abilities and difficulties, which could have limited the depth of the analyses. Nevertheless, this study provides new evidence regarding pragmatic processing in individuals with 22q11.2DS, although the underlying nature and extent of these alterations remain to be fully elucidated.

Data availability statement

The raw data supporting the conclusions of this article will be made available by the authors, without undue reservation.

Ethics statement

The studies involving humans were approved by Social Research Ethics Committee of UCLM under reference CAU-683200-X6H7. The studies were conducted in accordance with the local legislation and institutional requirements. The participants provided their written informed consent to participate in this study. Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article.

Author contributions

EM: Data curation, Project administration, Supervision, Writing – original draft, Writing – review & editing. NL: Writing – original draft, Writing – review & editing. PD-H: Writing – original draft, Writing – review & editing. PC-H: Writing – original draft, Writing – review & editing.

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The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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