

# Receptive and Expressive Vocabulary Performance in Young Children with 22q11.2 Deletion Syndrome

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## Abstract

**Aim:** This study investigated whether a discrepancy is found between the performances on receptive and expressive vocabulary of young children with 22q11.2DS, with the aim to contribute to the discussion in the literature about the relation between receptive and expressive language in children with 22q11.2DS.

**Methods:** 16 children with 22q11.2DS, ten girls and six boys, between the ages of 3;5 and 6;5 years participated in this study. Receptive vocabulary was measured using the Peabody Picture Vocabulary Test (PPVT) and expressive vocabulary was measured using the Active Vocabulary Test (AVT).

**Results:** No significant discrepancy was found between receptive and expressive vocabulary. Performances on both types of vocabulary were delayed compared to the norm group. The results indicate that young children with 22q11.2DS have delayed receptive and expressive vocabularies.

**Conclusions:** Early language treatment with the focus on both expressive and receptive vocabulary is necessary to reduce vocabulary delay and prevent problems with later educational outcomes such as verbal intelligence, reading skills and math skills.

## 1. Language impairment in 22q11.2DS

The 22q11.2 deletion syndrome is a genetic disorder caused by the deletion of a part of chromosome 22 (Antshel, Marrinan, Kates, Fremont, & Shprintzen, 2009). The 22q11.2 deletion syndrome (22q11.2DS from here on), formerly also known as DiGeorge or Velocardiofacial syndrome, can be inherited from parents, but in most cases the deletion occurs *de novo* (i.e., it appears for the first time in a new generation) (Persson et al., 2006; Rakonjac et al., 2016). Many individuals with 22q11.2DS experience psychiatric and physical problems, and have a developmental delay (Angkutsiri et al., 2014). The common physical anomalies are palatal defects like cleft palate, congenital heart diseases and hypoplasia. Since 1992, an elevated risk of developing schizophrenia and other related disorders has been described as a common feature in 22q11.2DS. Beside psychotic disorders, a wide range of developmental issues such as cognitive and temperamental delays has been found to be common among children with 22q11.2DS. Some of the most consistent problems that are found in children with the syndrome are Attention Deficit Hyperactivity Disorder (ADHD), Autism Spectrum Disorders (ASD) and anxiety disorders (Antshel et al., 2009). Moreover, low intellectual functioning is also widely observed in individuals with the syndrome.

Language, speech and learning problems are furthermore among the most common characteristics of 22q11.2DS. They are in fact, according to Golding-Kushner (2005), the problems that lead and should lead clinicians to suspect the presence of the genetic disorder. In a clinical environment, it can be crucial to make a distinction between language and speech problems for the right diagnosis and treatment. It is therefore important to also make a clear distinction between the definitions of speech and language in a research area with clinical purposes. Language here refers to the comprehension and formulation of ideas and concepts, while speech is the actual production of these concepts.

Golding-Kushner, Weller and Shprintzen (1985) were the first to describe the language profiles of children with 22q11.1DS aged 3-18 years old. They reported delays in language comprehension and delays in use of vocabulary, syntax and abstract reasoning in communicative situations. Later studies by Solot et al. (1998; 2000) showed that early language development was delayed in onset. Some studies even found that some of the children with 22q11.2DS were non-verbal until 30 months of age (Persson et al., 2006; Scherer, D'Antonio, & Kalbfleisch, 1999). In addition, Scherer et al. (1999) studied very young children with 22q11.2DS from 6-30 months of age and their results showed that the early vocabulary of the children was impaired in comparison to children with a combined cleft lip and palate or an isolated cleft palate of non-syndromic origin. There are many more studies that have found impairments in the language acquisition of children with 22q11.2DS (Moss et al., 1999; Solot et al., 2001; Glaser et al., 2002; Goorhuis-Brouwer, Kambanaros, Taxitari, Theodorou, Varnava, & Grohmann, 2018). These findings show that language should be an area of focus for clinicians and researchers who are working with 22q11.2DS, in order to help reduce delays in further language development of these children.

Results of most of the studies conducted with young children with 22q11.2DS show that children with 22q11.2DS have more severely impaired expressive language than receptive language (Golding-Kushner et al., 1985; Scherer et al., 1999; Solot et al., 2001; Goorhuis-Brouwer, Dikkers, Robinson, & Kerstjens-Frederikse, 2002; Persson et al., 2006; Van den Heuvel, Manders, Swillen, & Zink, 2018). There are some studies however, that find an expressive language advantage. To explain these (conflicting) advantages, implications about chronological age, mental age and physical anomalies are made in earlier studies. Still, a discussion on this subject remains. This study will also focus on the receptive-expressive language discrepancy in children with 22q11.2DS with the aim of contributing to this discussion.

## 2. Expressive and receptive language in TD children

Expressive language is the use of language (words, gestures, sentences) to express meaning. Skills that are important for expressive language are labelling objects in the world, using correct grammar and describing stories, actions and events. This is different from receptive language, which consists of the ability to understand language using linguistic, visual and auditory information (Fenson et al., 1994). In typically developing (TD) children, receptive language mostly emerges before expressive language (Fenson et al., 1994; Antshel et al., 2009). The first signs of word comprehension in typically developing children, such as reacting to their own name or names of their favourite people and/or toys, emerge around 9 months of age with spontaneously spoken words following around 12 months (Fenson et al. 1994). By the age of 12 months, however, infants have developed an understanding of many more words than at the age of 9 months. They can even understand short sentences but will not be able to produce any phrases of their own until between 18 and 24 months of age (Hudry et al. 2010). The development of language comprehension occurs ahead of production, because children cannot use words which they do not understand. Fenson et al. (1994) demonstrated the growth in comprehension over production skills in a study of infant language development, plotting parent-reported receptive and expressive vocabulary counts on the MacArthur-Bates Communication Development Inventory (MCDI; Fenson et al. 1992).

In 22q11.2DS, both typical and atypical discrepancies between expressive and receptive language are found (Van den Heuvel et al., 2018). A number of researches regarding receptive-expressive language skills in children with 22q11.2DS will be discussed below.

### 3. Expressive and receptive language in children with 22q11.2DS

#### 3.1 *Evidence for a receptive language advantage*

Solot et al. (2001) tested 53 preschool children with 22q11.2DS between the ages of 7 months and 2;6 years on language proficiency. They used a Pre-School Language Scale (PLS; Zimmerman, Steiner & Evatt, 2011) which measures early language development, tapping into a broad range of receptive and expressive language skills. The results of this test showed that in these children, expressive language skills often lagged behind receptive language skills. A quarter of these children scored significantly lower on expressive language and for the group as a whole, the expressive language scores were overall lower than the receptive skills scores.

Another research with young children with 22q11.2DS, using a different method, was done by Persson et al. (2006). They studied 19 preschool and school-aged children, between the ages of 4;11 and 8;5 years. To assess receptive vocabulary, the Peabody Picture Vocabulary Test (PPVT; Dunn & Dunn, 1997) was used. In this test, the child has to choose the correct picture from a set of four pictures in response to a target word that they heard. The children with 22q11.2DS scored low ( $M = 76$ ,  $SD = 15.25$ ) compared to the norm group ( $M = 100$ ,  $SD = 15$ ), suggesting difficulties with understanding words. These test results are similar to those found in earlier research by Golding-Kushner et al. (1985). To get insight in the expressive language abilities of the children, a Swedish version of The Bus Story test (BST) was used. In this test, the children are told a story while looking at pictures. Afterwards the child has to retell the story accompanied by the same pictures. The calculated expected information for the participants was -1.31 SD below the standardised mean score of  $PIQ = 100$ . This is lower than the normal-developing population mean. According to their results, expressive language was more impaired than receptive language. They also found a negative correlation between age and the information score of The Bus Story Test ( $r = -0.46$ ,  $p < 0.05$ ), implying that the older the children were, the more severe the problems in expressive language were. For receptive language, no such correlation was found. This suggests that the discrepancy continues in older ages.

This suggestion by Persson et al. (2006) has been supported by a research by Van den Heuvel et al. (2018). They tested 60 children with 22q11.2DS which were older than pre-schoolers, namely between 6 and 13 years. They tested expressive syntax and vocabulary using subtests of the Clinical Evaluation of Language Fundamentals (CELF-4; Semel et al., 2003) in which the children are asked to name a pictured object or action and recall sentences, and describe the relations between words, respectively. They tested receptive syntax using a CELF-4 subtest in which children are asked to follow oral directions and measured receptive vocabulary with the PPVT. The scores on these tests, in line with abovementioned studies, indicated a receptive language advantage for the children with 22q11.2DS. In this study, there were two testing moments. For both receptive and expressive language, progress was made between the first and the second testing moment. However, the receptive language advantage remained. The same results were found in Moss et al. (1999), who also used the CELF-4 with individuals with ages ranging from 6 to 27 years. This shows that the receptive language advantage persists as children get older.

#### 3.2 *Evidence for an expressive language advantage*

There are studies, however, that contradict the results that are discussed in the previous section. Glaser et al. (2002) also used the CELF, but an earlier edition, to assess school-aged participants with 22q11.2DS with ages between 6 and 19 years. In this CELF-3, roughly the same subtests are used as the ones described above to measure receptive and expressive language. The results of the tests

indicated lower receptive language skills than expressive language skills. However, Glaser et al. (2002) recognized that their results were not in line with previous results, due to age and task complexity differences across studies. Solot et al. (2001) which was discussed earlier, did not only test young children, but also studied a group of older children with 22q11.2DS with ages between 5;9 and 16;7 years. They similarly used a version of the CELF and also found an expressive language advantage, although a rather small one.

### *3.3 Hypotheses explaining a discrepancy in expressive and receptive language*

As the studies discussed above show, there are conflicting results with respect to the balance between the expressive and receptive language skills of children with 22q11.2DS. Most researchers who studied older children and found a receptive language advantage, also found that it persisted with age. The two studies that examined older children and found an expressive language advantage, also found this persistence. Not only chronological age, but also mental age can play a role in the discrepancies and therefore maybe also in the conflicting results. Due to the wide variability in mental age of children with 22q11.2DS, it remains difficult to define a clear-cut language profile for a group of children with 22q11.2DS. This causes differences in mental age within testing groups (Van den Heuvel et al., 2018). Due to this complexity, there is a limited number of studies comparing 22q11.2DS children with mental age matched controls, which prevents researchers from confirming this hypothesis.

Something that has been done research on and is controlled for in multiple studies, is physical anomalies. Since a large number of 22q11.2DS children have palatal defects, they are impaired in their speech development. A hypothesis is that this delayed development restricts the children in their use of expressive language. Scherer et al. (1999) compared the performance of children with 22q11.2DS with palatal deficits with that of children with a cleft lip/palate and an isolated cleft palate of non-syndromic origin. The development of these children shows several similarities with the development of 22q11.2DS children. Both populations have characteristics such as Velopharyngeal insufficiency (VPI), compensatory speech sounds and early language impairment. Scherer et al. (1999) used the Sequenced Inventory of Communicative Development-R to test expressive as well as receptive language of young children between 6 and 30 months. The results indicated that the 22q11.2 children showed more severe language impairments and a larger receptive vocabulary advantage compared to the cleft lip/palate and isolated cleft palate populations. These results indicate that palatal deficits do not have an effect on potential expressive language delays. The results also support the idea that the language impairment and the expressive-receptive discrepancy is distinctive to the syndrome, regardless of palatal anomalies.

### *3.4 The current study*

The current study was designed to address the following research question: Is there a discrepancy in the expressive and receptive vocabulary skills of young children with 22q11.2DS? The aim of this study is to contribute to the discussion in the literature about expressive-receptive language discrepancy in children with 22q11.2DS, as described in the section above. In order to make a valid comparison, both expressive and receptive vocabulary will be tested, reported and compared. This study is unique in that it only focuses on receptive and expressive vocabulary specifically. Most other studies focus on expressive and receptive language, which also cover syntax and morphology. The expressive and receptive language performance of young children with 22q11.2DS has not often been studied. The current study focuses on young children, including a relatively small age range in comparison with previous work, which might bring new insights to the discussion. This study will also help to better understand the language profile of this population, which can lead to improvements in the area of diagnostics and treatment of this syndrome.

Overall there are more studies that find evidence for a receptive language advantage in different ages, including very young children as in Scherer et al. (1999). There are only a few studies that found an expressive language advantage and therewith no convincing evidence. On the basis of these implications from earlier research, the expectation for this study is that the young children will show a receptive vocabulary advantage.

#### 4. Methods

The data that are used for this study are part of a larger longitudinal observational case-control study on language impairment in the 22q11.2 deletion syndrome, which is currently being conducted by principal investigator Frank Wijnen and colleagues. In this original study, the 22q11.2DS participant group will be compared to a group which consists of children with a Developmental Language Disorder and a group of typically developing children. This study is approved by the Medical Ethics Committee (METC). Since the study is still in an early phase, the sample size is dependent on the 22q11.2DS data that is currently available.

##### 4.1 Participants

In this study, 16 children with a confirmed genetic diagnosis of 22q11DS participated. Ten of these children are female and six are male. The ages of all participants fall within the range of 3;5 to 6;5 years ( $M = 62$  months,  $SD = 13$  months). The gender and precise age per participant are shown in Table 1. The participants are monolingual Dutch speaking children. All children have a 22q11.2DS diagnosis provided by a medical specialist. Four of the 16 children have a cleft palate deficit as reported by their parents. Children with a hearing loss of 35 dB or worse and children raised in a multilingual environment were excluded from the study. The participants were recruited at the Wilhelmina Kinderziekenhuis (WKZ), which has a 22q11.2DS expertise centre with a large cohort of patients. Participants were also recruited with help from the national parent support group *Stichting Steun 22q11*.

Table 1: *Descriptive data for the subjects*

Subject	Gender	Age (in months)	Cleft palate deficit
1	M	41	No
2	F	43	No
3	M	44	No
4	F	46	*
5	F	52	*
6	F	55	No
7	M	64	Yes
8	F	68	No
9	F	70	No
10	F	71	Yes
11	M	71	Yes
12	F	71	No
13	F	71	Yes
14	M	73	No
15	F	74	No
16	M	77	No

*\*This information has not yet been provided by the parents of the participant*

#### 4.2 Materials and Procedures

Parents were first informed about the research via the physician treating the child or via a message posted on the website of the *Stichting Steun 22q11*. If parents responded positively, they were contacted by a researcher who provided further information and conducted a short screening interview in order to assess if the participant met the inclusion criteria. If the inclusion criteria were met, they were asked to enrol in the study with their child and if the answer was positive, they signed an Informed Consent form. The parents were given multiple opportunities to ask questions about the research. The assessments were conducted in a child-friendly and informal setting at the school or day care of the participants.

To test receptive vocabulary, the PPVT-III-NL (Schlichting, 2005) was used. The PPVT is designed for an age range of 2;3 to 90;0 years. It consists of 17 sets of 12 items, which means the test contains 204 items in total. The sets are ordered according to difficulty. The PPVT-III-NL was administered and scored according to the official guidelines. During this test, the child is presented with single words, nouns and verbs, one at a time. The child is asked to match each word to one out of the four pictures presented by means of pointing. The duration of the task was approximately ten minutes.

The test that was used to measure expressive vocabulary is the Active Vocabulary Task (AVT), which is part of the CELF Preschool-2-NL (Wiig et al., 2012). The CELF Preschool-2-NL is an assessment that contains multiple subtests which together can diagnose and evaluate language problems. The assessment is designed for children between the age of 3;0 and 6;11 years. In this subtest, the child is presented with pictures of actions or objects, one at a time. The instructor tells the child to look at the picture and then asks the child to name what they see on the picture. The answers are assessed on the basis of scores; if the child answers correctly, two points are scored. If the answer is not as prescribed but is acceptable, one point is scored. If the answer is incorrect, 0 points are awarded to the child. If the child answers incorrectly or does not respond within 10 seconds, the instructor continues with the next picture. The duration of the task was approximately five minutes.

In addition to the procedures aimed at the children, all parents/caretakers were asked to complete questionnaires concerning physical deficits such as cleft palate deficits, socio-economic status, educational attainment, pedagogical style, and patterns of engagement in verbal activities (e.g. book reading, singing), which may affect children's language outcomes.

#### 4.3 Data analysis

For the PPVT, raw scores were converted to standard quotient scores ( $M = 100$ ,  $SD = 15$ ), using chronological age (CA) as the reference age. For the AV task, raw scores were first converted to scaled scores ( $M = 10$ ,  $SD = 3$ , range = 1-19). These scores were then used to calculate standard quotient scores ( $M = 100$ ,  $SD = 15$ ) using CA as reference age. Because a comparison within the same group was made, and the data were normally distributed, a Paired Samples T-Test was used to compare the children's performances on the PPVT and the AVT. For this statistical analysis, an  $\alpha$  of  $p < 0.05$  was the threshold for statistical significance. Normal distribution was tested using the Shapiro-Wilks Test (AV:  $p = 0.89$ , PPVT:  $p = 0.86$ ), because of the small sample ( $< 50$ ). The normal distribution was also confirmed by the skewness of 0.17 ( $SE = 0.56$ ) and kurtosis of -0.32 ( $SE = 1.10$ ) for the AVT and the skewness of 0.16 ( $SE = 0.56$ ) and kurtosis of -0.23 ( $SE = 1.10$ ) for the PPVT. The effect size will be measured using Cohen's  $d$  with the following rule of thumb by Cohen (1998): 0.2 = small, 0.5 = medium and 0.8 = large.

## 5. Results

### 5.1 The Peabody Picture Vocabulary Task

The mean standard quotient score of the PPVT for the 16 participants who performed this test was 81.6 (range = 55-112, SD = 15.47). Five of the 16 participants performed not more than 1 SD below the mean, compared with the CA norms. Three participants performed above the mean. Eight participants scored at least 1 SD below the mean (see Table 2).

Table 2: Raw scores and quotient scores for the PPVT and the AVT

Subject	Age (in months)	AVT Raw Scores	AVT Quotient scores	PPVT Raw Scores	PPVT Quotient scores
1	41	16	100	58	112
2	43	6	68	43	89
3	44	11	80	37	78
4	46	9	68	31	67
5	52	20	90	51	86
6	55	20	85	48	78
7	64	22	80	75	102
8	68	21	68	67	86
9	70	19	64	50	61
10	71	21	68	61	75
11	71	24	75	64	79
12	71	26	80	53	64
13	71	32	95	69	87
14	73	25	75	83	101
15	74	29	85	71	86
16	77	6	55	42	55

### 5.2 The Active Vocabulary Task

The mean standard quotient score of the AVT for the 16 participants who performed this test was 77.3 (range = 55-100, SD = 12). Compared with the CA norms, four of the 16 participants performed not more than 1 SD below the mean. None of the participants performed above the mean, one participant scored exactly the mean score, and eleven participants scored at least 1 SD below the mean (see Table 2).

### 5.3 Expressive-receptive vocabulary

A significant positive Pearson's correlation was found between the AVT and the PPVT ( $r = 0.632$ ,  $p < 0.01$  (2-tailed)). The mean quotient scores of the PPVT and the AVT (81.6 and 77.3 respectively) indicate that there seems to be a difference in performance on the two tasks. However, according to the Paired Samples T-Test, there is no significant difference between the children's performances on the PPVT and the AVT of the group ( $t(15) = -1.44$ ;  $p = 0.172$  (2-tailed)). The calculated Cohen's  $d$  is 0.36. The calculated Cohen's  $d$  indicates a small effect size ( $d < 0.5$ ).

## 6. Discussion

In the present study, 16 children with 22q11.2DS were tested on receptive and expressive vocabulary. This study investigated whether a discrepancy is found between the performances on receptive and expressive vocabulary of children with 22q11.2DS, with the aim to contribute to the discussion in the literature about the relation between receptive and expressive language in children with 22q11.2DS. The Peabody Picture Vocabulary Test and the Active Vocabulary Test were conducted with the children, in order to test receptive and expressive vocabulary respectively. The results show that there is no significant difference in performance of young children with 22q11.2DS on receptive and expressive vocabulary tests. This is contradictory to the hypothesis of this study, which was that a receptive vocabulary advantage would be found. The results will be further discussed in this section, together with suggestions for further research.

The quotient scores in Table 2 indicate that the children with 22q11.2DS have impairments in both expressive and receptive vocabulary. For expressive vocabulary, all children except one scored below the standardised mean score of the TD population. Three participants scored average (between +1 and -1 SD). Thirteen participants scored below average, of which five participants scored little below average (between -1 and -1.5 SD), two participants scored clearly below average (between -1.5 and -2 SD below), and six scored much lower than average (-2 SD and lower). The average mean of the group for the AVT is 77.3, which falls in the category clearly below average. This classification of scores is based on the testing manual of the CELF-4 (Wiig et al., 2012).

For receptive vocabulary, the quotient scores are subtly higher than those on the AVT, but this difference is not significant. Eight participants scored average (between +1 and -1 SD). Eight participants scored below average, of which three participants scored little below average (between -1 and -1.5 SD), one participant scored clearly below average (between -1.5 and -2 SD below), and four scored much lower than average (-2 SD and lower). The average mean of the group for the PPVT is 81.6, which falls in the category little below average. This classification of scores is also based on the testing manual of the CELF-4 (Wiig et al., 2012). Thus, no significant receptive or expressive vocabulary advantage was found, but the data merely point out that both types of vocabulary are weaker in comparison with the norm group.

The information that can be derived from the quotient scores shows that young children with 22q11.2DS have a delay in vocabulary acquisition. This can be problematic for later development. According to multiple studies with TD children, both receptive and expressive vocabulary performance in young children are predictors of childhood outcomes like verbal intelligence, reading ability and math skills in later development. Fewell & Deutscher (2002) investigated if performance on receptive vocabulary in a large sample of young TD children (N = 543) at 36 months of age would predict verbal ability and reading at 5 and 8 years of age. They used the PPVT to test receptive vocabulary. The Wechsler Preschool and Primary Scale of Intelligence was used to assess verbal IQ at the age of 5 years and the Wechsler Intelligence Scale for Children at the age of 8 years. The Woodcock-Johnson Tests of Achievement tested reading ability of the children at the age of 8 years. Three separate regression analyses were used to test the contributions of receptive vocabulary on the Verbal IQ measures at ages 5 and 8 and on reading at age 8. In all three models, children's receptive vocabulary quotient scores at the age of 36 months accounted for a large portion of variance of the outcome variables of verbal IQ measured at 5 and 8 years (37% and 33% respectively) and of reading at age 8 years (19%). Another longitudinal study that found that receptive vocabulary predicts childhood outcomes, was conducted by Sénéchal and Lefevre (2002). They used a similar regression model as in Fewell & Deutscher (2002) to examine the long-term links between receptive vocabulary (PPVT) and reading achievement in third grade. The results indicated that children's reading skills in grade 3 were predicted by children's early receptive language skills. In addition,

Bleses, Makransky, Dale, Højen and Aktürk Ari (2015) conducted a longitudinal study of 10 years with a large sample of Danish TD children, starting with 16 to 30 months old children. Expressive vocabulary skills were assessed using the Danish adaptation of the Child Development Inventory (CDI). Reading and math skills were assessed using The Danish National Tests, which are administered at the end of every school year. They found that early expressive vocabulary was predictive of later reading and math outcomes, in particular decoding and reading comprehension, in sixth grade. They also found that low scores have the greatest predictive power, indicating that children with early delays have elevated risk for later reading problems.

The studies reported above provide evidence for a negative effect of early vocabulary delay on educational outcomes in later development. It is therefore important that young children with 22q11.2DS, who were found to have a vocabulary delay in the current study, receive early language treatment to prevent problems with reading, verbal ability and mathematic skills. This treatment should be focused on receptive as well as expressive vocabulary, as the current study shows that children with 22q11.2DS have difficulties in both domains.

The positive correlation between the AVT and the PPVT with a moderate strength of association ( $r > 0.5$ ; Hinkle, Wiersma & Jurs, 2003) indicates that the better a child performs on receptive vocabulary, the better (s)he also performs on expressive vocabulary and the other way around. If the children get language therapy on both expressive and receptive vocabulary at the same pace and on the same level of difficulty, the two types of vocabulary may enhance each other, resulting in a better vocabulary performance. Besides that, the correlation does not have a large strength of association, which means a delay in expressive or receptive vocabulary cannot be ruled out if one of the two types of vocabulary does not get as much attention in language therapy as the other type. This confirms the idea that focus in language therapy for young children with 22q11.2DS should not be on either expressive or receptive vocabulary, but on both.

The expectation that was reported earlier in the present study, was made on the basis of findings from previous studies. Most studies showed evidence, and also most convincing evidence, for a significant receptive language advantage. A reason for the difference between the findings of these studies and the current study could be a methodological matter. Most of these studies tested their participants on receptive and expressive language, covering expressive and receptive vocabulary, morphology and syntax. The current study only focused on vocabulary. It is therefore possible that the receptive advantage is primarily carried by syntax and/or morphology. In order to confirm this hypothesis and to gain more information on the expressive-receptive language discrepancy, further research should focus on other language aspects. This can be done by assessing receptive and expressive syntax and morphology in separate tests with children with 22q11.2DS and directly comparing them to each other, as is done in this study with vocabulary.

Another factor that might explain the difference between the findings of earlier studies and the current study, is the sample size ( $N = 16$ ). This sample is smaller compared to samples that are used in earlier studies (Solot et al., 2001, Van den Heuvel et al., 2018, Moss et al., 1999), although not much smaller. As the effect size ( $d = 0.36$ ) reported earlier also indicates, a larger sample may yield a significant but small receptive vocabulary advantage in young children with 22q11.2DS. It is therefore important for further research to include large sample sizes, as in the longitudinal study with 22q11.2DS children that is currently being conducted by principal investigator Frank Wijnen and colleagues.

In addition, the age of the participants could play a role in the dissimilarity between the outcomes of previous studies and the current study. This study included young children between 3;5 and 6;5 years of age. Since most previous studies focused on school-aged children and adolescents, it could be that a significant receptive language advantage is found in children of an older age and not in pre-school children. An explanation for this assumption might be that a receptive language

advantage is a typical development also for TD pre-school children, with whom the children with 22q11DS are compared to. TD school-aged children however, might have found a balance between receptive and expressive language, while school-aged children with 22q11.2DS have not. This way, a comparison of school-aged children with 22q11DS with TD school-aged children might show a significant receptive language advantage.

More research should focus on language in children with 22q11.2DS in the early ages. Not only to attain more knowledge about the relation between age and expressive and receptive language in this target group, but also because, as mentioned earlier, research indicates that very early vocabulary delays negatively affect later educational outcomes. In order to prevent these problems, language therapy for young children should be available and appropriately adjusted to their language abilities. To attain this goal, more knowledge about language abilities in the early ages of children with 22q11.2DS is necessary.

## 7. Conclusions

Young children with 22q11.2DS between the ages of 3;5 and 6;5 years were tested on their receptive and expressive vocabulary. Results indicate no significant discrepancy between expressive and receptive vocabulary. However, both types of vocabulary are delayed. This implicates that language therapy should focus on both receptive and expressive vocabulary at the same level of difficulty for 22q11.2DS children of a young age. In addition, this language therapy for young children should be available at a young age and appropriately adjusted to their language abilities, in order to prevent problems in later educational outcomes.

Furthermore, further research should focus on acquiring more knowledge about a possible receptive-expressive language discrepancy by studying language aspects such as syntax and morphology in an isolated manner, using larger samples which also include very young children, and by conducting longitudinal studies.

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