LONGITUDINAL TRACKING OF VOCABULARY DEVELOPMENT IN CHILDREN WITH NEURODEVELOPMENTAL AND GENETIC DISORDERS USING COMMUNICATIVE DEVELOPMENT INVENTORIES (CDIs)

ZUZANNA LAUDAŃSKA [1,2], PATRICE VAN DER VENNE[1], HELENA PREIS[1], CLARA HOCHREITER[1], PAULINE KRIEGER[1], STEFFI SACHSE[3], LUISE POUSTKA[1], DAJIE ZHANG[1,4,], PETER B MARSCHIK[1,4,5,6,]

[1]Department of Child and Adolescent Psychiatry and Psychotherapy, Systemic Ethology and Developmental Science, Heidelberg University Hospital, Heidelberg University, and German Center for Mental Health (DZPG), Heidelberg, Germany

[2] Institute of Psychology, Polish Academy of Sciences, Warsaw, Poland

[3] Institute of Psychology, Heidelberg University of Education, Heidelberg, Germany

[4] iDN – interdisciplinary Developmental Neuroscience, Department of Neurology, Medical University of Graz, Austria

[5]Center of Neurodevelopmental Disorders (KIND), Department of Women's and Children's Health, Centre for Psychiatry Research, Karolinska Institutet and Region Stockholm, Stockholm, Sweden

[6]Child and Adolescent Psychiatry and Psychotherapy, University Medical Center Göttingen and German Center for Child and Adolescent Health (DZKJ), Göttingen, Germany

Corresponding author: Peter B. Marschik, E-mail**:** Peter.Marschik@med.uni-heidelberg.de

Received: 15.10.2025
Accepted: 02.01.2026

REVIEW ARTICLE
UDK: 81'23-056.36-056.7
DOI: https://doi.org/10.31299/hrri.62.si.11

Abstract: *MacArthur-Bates Communicative Development Inventories (CDIs) are widely used to assess early language development and identify language delay in children, including those with neurodevelopmental and genetic conditions. This vignette reviews studies that used repeated CDI assessments to track expressive vocabulary growth over time. We identified 13 longitudinal studies including participants with autism, Down syndrome, Williams syndrome, and other rare genetic conditions. The results show that, across different conditions, expressive vocabulary development was delayed and highly heterogeneous, but few studies included more than two time points, hence limiting inferences about non-linear trajectories, plateaus, or regression. The review highlights both the potential and current limitations of CDI-based longitudinal data and underscores the need for harmonised, multi-timepoint study designs in order to better characterise expressive language development in atypical populations.*

Keywords: *Autism, Down syndrome, Williams syndrome, Angelman syndrome, 5p-deletion syndrome, vocabulary, language, Communicative Development Inventories*

INTRODUCTION

Communicative Development Inventories (CDIs) have transformed the study of early language and communicative development since the mid-1990s, offering insights into the acquisition of gestures, vocabulary, grammar, and communication from infancy onward (Fenson et al., 1994). In addition to characterising typical development, CDIs have been (a) used to detect "red flags" for language delay (e.g., Guinchat et al., 2012; Herlihy et al., 2015), and (b) applied in studies of children with neurodevelopmental conditions and genetic syndromes, often beyond the age range for which the CDIs were designed. These studies encompass clinical screening, developmental monitoring, outcome prediction, as well as the exploration of condition-specific developmental profiles (e.g., Luyster et al., 2007; Tager-Flusberg et al., 2009; Laudańska et al., 2026). In this vignette, we focus on a specific subset of CDI applications in clinical populations: studies using longitudinal designs with repeated CDI assessments to track expressive vocabulary growth over time. Longitudinal CDI data can distinguish delayed, yet progressing vocabulary growth from plateauing growth or stagnation – patterns that are undetectable in single-timepoint assessments (e.g., Marschik et al., 2007).

METHODS

This analysis draws on our systematic review of expressive vocabulary development in infants with neurodevelopmental conditions and genetic syndromes (Laudańska et al., 2026). For the present analysis, we extracted relevant information to provide descriptive reports on studies with longitudinal designs that (a) report multiple CDI assessments at different time points, and (b) provide raw scores for expressive vocabulary.

RESULTS

We identified 13 longitudinal studies using CDIs to track expressive vocabulary in children with neurodevelopmental or genetic conditions (see Supplementary Table): 5 studies on autism, 4 on Down syndrome (DS), and 1 on Williams syndrome (WS), as well as 2 case reports on 5p-deletion syndrome and 1 on Angelman syndrome. With respect to language and geographic context, English-speaking children represent the largest group studied, though publications also included Italian-, Dutch-, Mandarin-, Farsi-, and Norwegian-speaking samples. Figure 1 presents condition-specific age trends (raw mean scores for expressive vocabulary, weighted to account for differences in sample sizes) for autism, DS, and WS. These visualisations underscore the variability in developmental pace and trajectory across different syndromes. In the case reports using CDIs at multiple time points, Kristoffersen (2020) and Kristoffersen & Simonsen (2025) examined lexical and gestural development in 5p-deletion syndrome between 24 and 87 months (Fig. 2; case studies with 3, 14, and 20 timepoints). Han et al. (2019) used CDIs in a pharmacological supply study of twins with Angelman syndrome, conducting 3 repeated assessments in 6-month intervals. Twins A and B were enrolled at 101 months, twin C at 68 months, and twin D at 84 months. All four participants showed no growth in expressive vocabulary over the observation period.

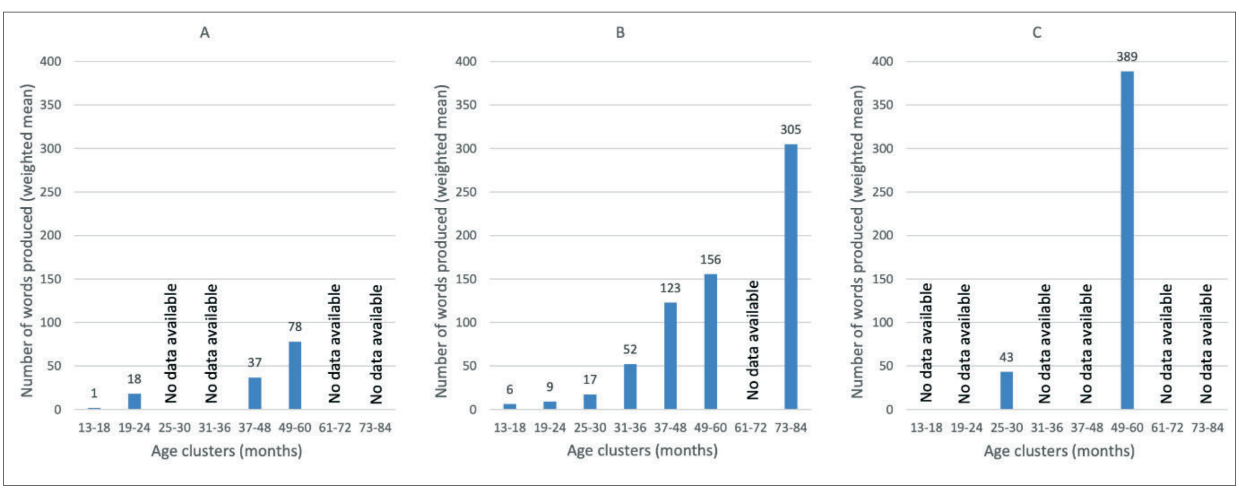

Figure 1. *Expressive vocabulary based on raw mean CDI scores (weighted by sample size) derived from longitudinal assessments and grouped into age clusters for autism (A), Down syndrome (B), and Williams syndrome (C).*

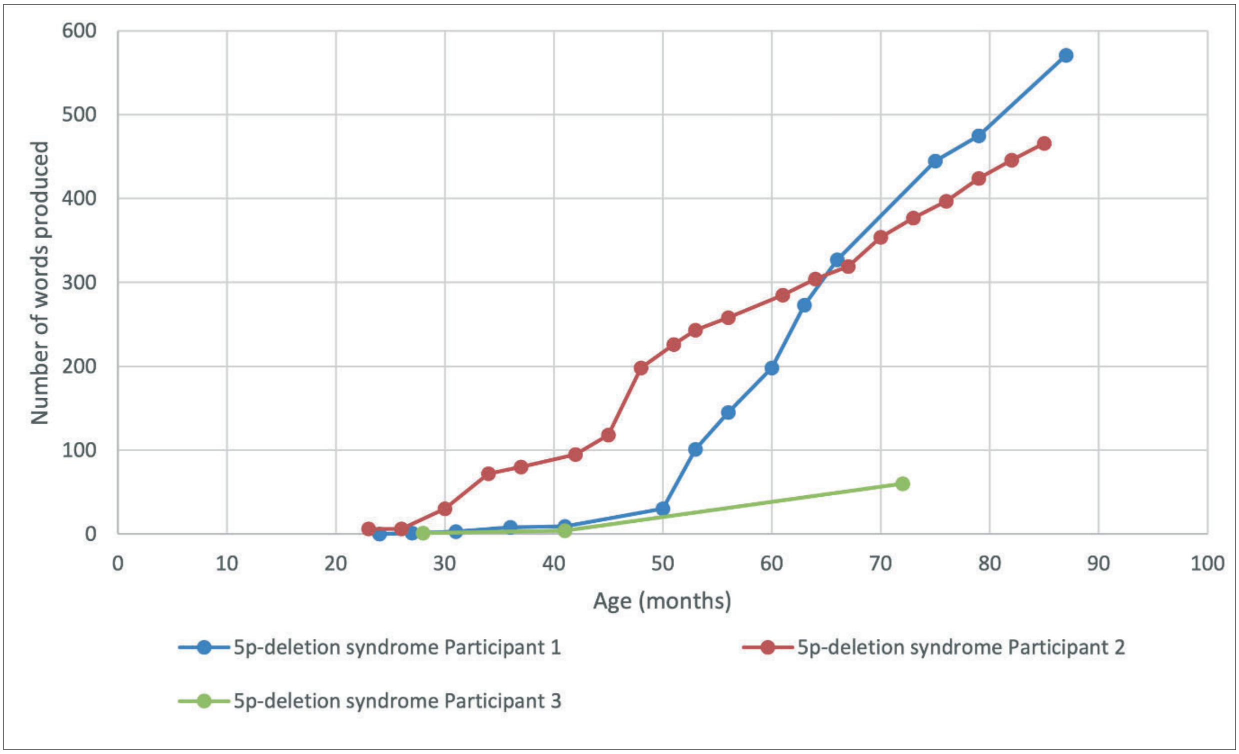

Figure 2. *Trajectories of expressive vocabulary in 5p-deletion syndrome based on three longitudinal case studies (Kristoffersen, 2020; Kristoffersen & Simonsen, 2025).*

DISCUSSION

This article reviewed longitudinal applications of CDIs to assess expressive vocabulary development in children with neurodevelopmental and genetic conditions. The aim was to examine whether existing longitudinal CDI data allow for the identification of vocabulary growth patterns and variability. While the available evidence suggests delayed onset and substantial heterogeneity, data limitations (fewer time points, small samples, and heterogeneous methods) preclude firm conclusions about condition-specific growth trajectories.

Out of the 50 studies using CDIs in autism research that were identified in our systematic review (Laudańska et al., 2026), only five had multiple CDI assessments spanning from 10 to 84 months of age. Compared to typically developing (TD) children, autistic individuals showed delayed expressive vocabulary development and highly heterogeneous developmental trajectories. However, due to limitations in data reporting, we could not extract specific profiles or document regression, as frequently reported in autism (Boterberg et al., 2019; Ozonoff & Iosif, 2019). To document specific trajectories and the loss of previously acquired functions, adaptations of existing study designs are warranted (e.g., Zhang et al., 2019). Especially in the field of autism, repeated measures to report transient or consistent language acquisition phenomena (e.g., Marschik et al., 2007) bear the potential to contribute to a better understanding of disorder-specific or neurodiverse pathways. In the context of DS, 4 studies reported longitudinal assessments of expressive vocabulary development. Since gesture use is considered a strength in children with DS, some authors (e.g., Deckers et al., 2016) chose to include both spoken and signed words as measures of expressive vocabulary. In addition, these studies included participants between the ages of 11 and 105 months - age ranges that are typically beyond the CDI design – thus, accounting for delayed language development in DS (e.g., Berglund et al., 2001). Only 1 publication on WS had a longitudinal design, also revealing a delayed onset of language production. Becerra and Mervis (2019) followed a larger group of children with WS over a longer period documenting pathways from pre-linguistic development and gestures to later speech-language outcomes. This highlights again that study designs would need to be adapted to decipher specific trajectories. Information on vocabulary development in 5p-deletion syndrome could be extracted from 2 case studies: all participants showed a delayed, yet growing expressive vocabulary, although to different extents and at different ages (Kristoffersen, 2020; Kristoffersen & Simonsen, 2025). Interestingly, no vocabulary growth could be shown for the 2 twin pairs with AS studied by Han et al. (2019). However, other researchers have suggested that phenotypes differ and individuals with non-deletion AS have higher communicative intent (Jollef et al., 2006).

Among all the studies reviewed, only 4 papers (of which 3 were case reports) included more than two timepoints. This is in stark contrast to research in typically developing populations, where ≥ 3 measurement points are common (e.g., Marschik et al., 2007; Peter et al., 2019). While two-timepoint models may capture group-level trends or intervention effects, they are insufficient to model individual growth trajectories or detect non-linear changes (Parsons & McCormick, 2024). Multi-timepoint designs are required to track periods of acceleration, stagnation, or regression, as well as to thoroughly analyse vocabulary growth curves in children with neurodevelopmental disorders and genetic syndromes. Further, children with elevated likelihood for developmental language disorders should be studied in prospective, longitudinal frameworks to better understand risk and resilience factors. Additional challenges across studies include small sample sizes, inconsistent administration procedures, and heterogeneous reporting formats. One method involves converting raw scores to proportions (e.g., Belteki et al., 2025), though this requires transparent reporting and ideally open repositories such as Wordbank (Frank et al., 2021).

CONCLUSION

In summary, existing CDI-based longitudinal research in neurodevelopmental and genetic conditions remains sparse and fragmented, falling short of delineating condition-specific developmental profiles, which is a long-standing aim of developmental clinical research. To advance our understanding of early expressive language development in these populations, future research should prioritise harmonised, multi-timepoint study designs, scalable digital implementation, open data practices, and standardised reporting.

Acknowledgment

This work was supported by the Deutsche Forschungsgemeinschaft (DFG, German Research Foundation) Grant Nr. 456967546 (PM, LP) and Grant Nr. 454648639 - SFB 1528: Cognition of Interaction C03 (PM); the Volkswagen Foundation project IDENTIFIED (PM); the National Science Center of Poland Grant 2022/45/N/HS6/02085 (ZL); the German Center for Mental Health project DZPG VISIONS (ZL), and the German Center for Child and Adolescent Health (DZKJ). This study has been discussed within the framework of the Heidelberg Flagship Initiative Family Transitions.

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