


## REVIEW ARTICLE

# Neurodevelopmental disorders in children with 22q11.2 deletion syndrome and recommendations for pediatric follow-up

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**Summary**

Neurodevelopmental disorders are the most prevalent chronic diagnoses in pediatric primary care, with rising incidence and significant impact on cognitive, motor, social, and communication functioning. 22q11.2 deletion syndrome (22q11.2 DS)—the most common human microdeletion syndrome—presents with a broad spectrum of somatic and neurodevelopmental abnormalities. Nearly all individuals with 22q11.2 DS show neurodevelopmental difficulties, including delays in motor and speech milestones, cognitive impairments, and behavioral challenges. The disorder affects approximately 1 in 2,500 newborns and is also associated with congenital heart defects, palatal anomalies, hypocalcemia, and immunodeficiency. Neurodevelopmental manifestations typically begin in infancy with delayed motor and speech development and progress into school age with difficulties in learning, attention, and peer interaction. Intellectual disabilities are common, with a distribution skewed toward lower IQ scores. Children often exhibit a verbal-performance IQ discrepancy and may experience further cognitive decline in adolescence or adulthood. Over 40% of affected individuals meet criteria for autism spectrum disorder, attention-deficit/hyperactivity disorder, or both. They also have increased risks for psychiatric conditions such as anxiety, depression, and schizophrenia. Pediatricians, as primary care providers, play a critical role in early identification and long-term monitoring. Recommendations include routine developmental assessments, early interventions (e.g., speech and occupational therapy), and regular IQ and adaptive functioning evaluations, especially during educational transitions. Early diagnosis and individualized, multidisciplinary approaches are essential to improve developmental outcomes and quality of life in children with 22q11.2 DS.

**Keywords:** 22q11.2 deletion syndrome, DiGeorge syndrome, neurodevelopmental disorders

## INTRODUCTION

Neurodevelopmental disorders arise during childhood, are usually multifactorial, and create chronic, often lifelong cognitive, motor, social, speech, communication, and other problems. This definition excludes disorders such as anxiety or depression in children and adolescents, which have a more episodic occurrence. In addition, ICD-11 classification excludes psychoses (e.g., schizophrenia) and bipolar affective disorder (1). Developmental problems are the most common chronic diagnosis made in primary pediatric health care. Their prevalence is not only high, but also increasing and reaches 17.08%, with the most significant contribution of attention-deficit/hyperactivity disorder (9.5%), autism spectrum disorder (2.5%) and intellectual disability (0.9%-1.2%,  $P < .05$ ). Only the prevalence (4.1%) of disorders classified as other developmental delay seems to be slightly decreasing (2). Research studies also show that developmental disorders are more common among boys, children with birth weight  $\geq 2500$  g, those of lower socioeconomic status, and those who live in urban areas, with less educated mothers or with a mentally ill family member (2,3).

A term 22q11.2 deletion syndrome (22q11.2 DS) is a collective name for a range of phenotypes previously described as DiGeorge syndrome, Shprintzen or Velocardiofacial syndrome, or Conotruncal anomaly face syndrome. It is caused by a deletion of the 22q11.2 region of about 2.5-3 Mb in size, which is the most common (approximately 1:2500 newborns) microdeletion occurring in the human genome. 22q11.2 DS is characterized by a wide range of neurodevelopmental and somatic imbalances, growth disturbances, anomalies of the heart, palate, and urinary tract, hypoparathyroidism with hypocalcemia, and thymic hypoplasia with T-cell immunodeficiency as presented in **Table 1** (4-8). Neurodevelopmental symptomatology can be observed in almost all children with 22q11.2 DS by careful clinical examination and/or formal testing (4).

## METHODS

For this narrative review, a comprehensive literature search was conducted using the PubMed database. The search included studies published at any time up to March 2025, without restrictions on publication date. The following keywords were used individually and in combination to identify relevant references: 22q11.2 deletion syndrome, neurodevelopmental disorders, and follow-up. The selection of articles focused on studies addressing the neurodevelopmental outcomes and follow-up of individuals with 22q11.2 deletion syndrome, including both clinical and research-based findings. Additional references were identified through manual screening of citations within the selected articles to ensure a broad and

representative overview of the available literature. Only English-language publications were considered.

## DIAGNOSIS OF 22q11.2 DS

The first step in the diagnosis of 22q11.2 DS is to raise the clinical suspicion of this syndrome, or, in less clear phenotypic presentations, of a genetic dysmorphic disorder in general (9,10). After that, during pre-test counseling, we discuss with patients and/or parents the potential benefits and limitations of genetic testing and choose the most appropriate test. Test results will be discussed during post-test counseling.

The detection of microdeletion 22q11.2 in the genome of affected children and adults has become much easier and more precise with the introduction of modern genomic testing (11). As presented in **Table 1**, the gold standard is molecular karyotyping (chromosomal microarray) on a sample of the patient's extracted DNA. This analysis enables the detection of microdeletion, but also precise characterization by determining its size and exact localization in the genome. Moreover, this approach allows, with very high confidence, searching throughout the genome and exclusion of associated chromosomal rearrangements (4).

Next-generation sequencing (NGS) is a cutting-edge diagnostic approach in genetics that has changed the face of clinical genetics over the last decade. Initially designed for the detection of intragenic variants (e.g., point mutations), this method has now been perfected to detect also chromosomal variants (aberrations) with a high precision that is only slightly lower than the precision of molecular karyotyping. Thus, in patients with an unclear phenotype, which is so common in the clinical geneticist's practice, this single method can provide a comprehensive genome search, including the detection of both intragenic and chromosomal variants (11,12).

More traditional diagnostic approaches (e.g., fluorescence in situ hybridization or multiplex ligation-dependent probe amplification (MLPA)) are usually used when the patient's phenotype is clear. Therefore, genomic search is not necessary (9). Additionally, MLPA, as a fast and not overly expensive method, can be used for a wide range of testing in a less selected group of patients (13).

## SPECTRUM OF NEURODEVELOPMENTAL DISORDERS IN 22q11.2 DS

Individuals with 22q11.2 DS are at increased risk for neurodevelopmental disorders, specifically for a range of cognitive, behavioral, and emotional difficulties. This can result in various problems in the sphere of learning, interpersonal relationships, and social functioning (14). In this article, we will briefly review the most common

**Table 1.** Basic principles of management of patients with 22q11.2 deletion syndrome

| Management category  | Key Points   |
|--|--|
| When to suspect 22q11.2 deletion syndrome                        | <ul style="list-style-type: none"> <li>• congenital heart defects (especially conotruncal anomalies)</li> <li>• hypocalcemia/hypoparathyroidism</li> <li>• immune deficiency (recurrent infections)</li> <li>• facial dysmorphism, palatal anomalies</li> <li>• developmental delay (speech, motor, social)</li> <li>• behavioral issues (ADHD, ASD, anxiety)</li> </ul>   |
| Recommended diagnostic genetic tests                             | <ul style="list-style-type: none"> <li>• First-line: CMA; consider MLPA or FISH if phenotype is typical</li> <li>• second-line: NGS if CMA is negative, for broader (intragenic) variant detection</li> </ul>  |
| Basic principles of treatment (other than developmental support) | <ul style="list-style-type: none"> <li>• specialist-directed treatments: management of cardiac, palate, renal, autoimmune, endocrine, and psychiatric conditions is guided by respective specialists</li> <li>• immune system support: aggressive infection management, possible use of prophylactic antibiotics or immunoglobulins; consider irradiated blood products/postponement of live vaccines until immune function normalizes</li> <li>• growth, nutritional, and gastrointestinal care: consider growth hormone, calcium supplementation, treatment for reflux or dysmotility disturbances</li> <li>• sensory support: hearing aids for hearing loss; treatment for ocular anomalies.</li> </ul> |
| Prenatal testing recommendations                                 | <ul style="list-style-type: none"> <li>• testing of parents before pregnancy for an accurate assessment of recurrence risk</li> <li>• CMA is the most commonly used method, which also analyzes other chromosomal regions</li> </ul>   |

ADHD – attention deficit hyperactivity disorder; ASD – autism spectrum disorder; CMA – chromosomal microarray; FISH – fluorescence in situ hybridization; MLPA – multiplex ligation-dependent probe amplification; NGS – next generation sequencing

neurodevelopmental concerns encountered in children and adolescents with 22q11.2 DS.

In infancy and toddlerhood, the main concerns are related to taking care of heart defects, feeding problems, acquiring milestones of gross motor skills, first words, and basic social skills in children with 22q11.2 DS. In general, the mean age at walking is 18 months, associated with a delay in the acquisition of speech (many are non-verbal until the age of 2-3 years, often with an acceleration in expressive speech after that) (15). A recent study confirmed that children with 22q11.2 microdeletion have impaired articulation skills and expressive language abilities. However, it did not report weaker receptive language skills and immediate verbal memory compared to healthy controls. Authors propose that children with 22q11.2DS should be considered as a risk category for the development of speech–sound pathology and expressive language abilities, and should be examined and included in a program of early speech–language stimulation immediately after diagnosis (16).

As the start of school approaches, intellectual disability, difficulties in learning and attention, as well as functional relationships with peers, come into focus. For early recognition, it is helpful to know that children and adults diagnosed with intellectual disabilities are mainly recruited from the population of the youngest with a diagnosis of global developmental delay, and this is the case not only for 22q11.2 DS but also for other syndromes with developmental challenges. Children and youth with 22q11.2 have a normal distribution of intellectual achievement shifted by about 30 points to the left compared to the general population, and with peak incidence at IQ 70-75. The incidence further declines on both sides, with about 40-45% of those having mild to moderate in-

tellectual disabilities (17,18). It is worth mentioning that a relatively significant proportion of people with 22q11.2 DS face a relatively mild but gradual intellectual decline after elementary school age and later during adulthood (19,20). Also, it should be noted that the total IQ score does not accurately represent the functionality of an affected individual, given the significantly higher verbal IQ score compared to the performance IQ score. Thus, these two dimensions should be considered separately (15).

In the study of Niklasson et al, 100 consecutively referred individuals with 22q11.2 DS were investigated for autism spectrum disorders (ASDs), attention deficit/hyperactivity disorder (ADHD), and intellectual disability (ID) using in-depth neuropsychiatric assessments and questionnaire screens. ASDs, ADHD, and ID were diagnosed in 23, 30, and 51 individuals, respectively, with some patients having more than one of these diagnoses. Females had a higher IQ than males. The results show that more than 40% of individuals with 22q11.2 DS meet criteria for either ASD, ADHD, or both (18). It also seems that individuals with 22q11.2 microdeletion, as well as with some other CNVs, who don't meet full autism diagnostic criteria, have elevated levels of autistic traits (21).

The risk of a child developing some of the neuropsychiatric disorders later during adolescence and life is increased, and is also among the major concerns of parents. Early predictors might involve attention deficit, hyperactivity, poorer impulse control, shyness, withdrawal, and autistic features with or without intellectual or language impairment. Current data show that individuals with 22q11.2 DS are at increased risk for anxiety, depression, and psychosis, with schizophrenia being diagnosed in approximately 25% of patients (22).

## RECOMMENDATIONS FOR SCREENING OF NEURODEVELOPMENTAL DISORDERS IN 22q11.2 DS FOR PEDIATRICIANS

The task of the health and other related systems is to recognize the initial phase of neurodevelopmental delay on time and, through informing, supporting, and specific programs, enable patients and parents to mitigate the consequences for the functionality and the quality of life as a whole. It is helpful to know that intellectual and overall cognitive achievements do not only depend on genetic factors, i.e., microdeletion and modifier genes, but also on parental, family, and broader social factors of resilience and vulnerability. This, together with the increased risk for behavioral and psychiatric problems, suggests the need for early psychological/psychiatric monitoring and interventions in individuals with 22q11.2 DS (17).

Pediatricians are professionals who are the first to meet children and follow them, in many countries, until the end of secondary school or up to 18 years of age. Further in this section, we present some of the recommendations for developmental assessment and follow-up of children with 22q11.2 DS that can be useful to pediatricians at different levels of health care, especially primary care:

- Formal developmental testing is recommended for children of all ages at the time of diagnosis.
- In case of early diagnosis of 22q11.2 DS, formal testing is recommended as early as the first year of life, even without clinically apparent developmental delays. Almost all infants/children with the syndrome show at least some degree of neurodevelopmental delay.
- For infants and toddlers, early consideration and implementation of developmental interventions is necessary and usually includes physio/occupational/speech therapy and sensory integration.
- The assessment of speech and communication should include not only expressive speech, but also receptive speech (i.e., comprehension); otherwise, it can lead to overestimation of a child's capacities.
- For school-age children, periodic formal assessments of IQ and adaptive functioning are recommended, es-

pecially during transition periods (preschool-primary and elementary-secondary school).

- The type of schooling depends on overall cognitive capacities, educational profile, and various individual and environmental factors. Schooling with adapted educational programs, as well as more intensive interventions (supports) in the educational process, is considered (15,23).

## CONCLUSION

Neurodevelopmental disorders are present in the vast majority of pediatric patients with 22q11.2 DS. As children grow older, the prevalence of specific neurodevelopmental entities changes, along with possible cognitive decline and the occurrence of neuropsychiatric disorders in some affected individuals.

Repeated formal neurodevelopmental assessments from early childhood to adolescence are recommended to provide better support, optimize interventions, and generally promote better health and quality of life. Although most of the recommendations listed above are applicable to everyone with 22q11.2 DS, management of developmental issues can be tailored to the characteristics of the individuals and the capacities of the community. Hence, some patients may require an even more comprehensive approach to the recognition and monitoring of neurodevelopmental risks/disorders.

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## NEURORAZVOJNI POREMEĆAJI KOD DECE SA 22q11.2 DELECIJSKIM SINDROMOM I PREPORUKE ZA PEDIJATRIJSKO PRAĆENJE

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### Sažetak

Neurorazvojni poremećaji predstavljaju najčešće hronične dijagnoze u primarnoj pedijatrijskoj zaštiti, sa sve većom učestalošću i značajnim uticajem na kognitivno, motoričko i socijalno funkcionisanje. Sindrom delecije 22q11.2 (22q11.2 DS) — najčešći mikrodelecijski sindrom kod ljudi — karakteriše širok spektar somatskih i neurorazvojnih abnormalnosti. Gotovo svi pojedinci sa 22q11.2 DS pokazuju neurorazvojne poteškoće, uključujući kašnjenje u razvoju motorike i govora, kognitivna oštećenja i probleme u komunikaciji i ponašanju. Ovaj poremećaj pogađa otprilike jedno od 2.500 novorođenčadi i povezan je i sa urođenim srčanim manama, anomalijama nepca, hipokalcemijom i imunodeficijencijom. Neurorazvojne manifestacije najčešće počinju u ranom detinjstvu kašnjenjem u razvoju motorike i govora, a nastavljaju se u školskom uzrastu kroz poteškoće u učenju, pažnji i socijalnim interakcijama. Intelektualne teškoće su česte, sa distribucijom učestalosti pomećenom ka ni-

žim vrednostima koeficijenta inteligencije. Deca često pokazuju razliku između verbalnog i neverbalnog skora i mogu imati dodatni kognitivni pad tokom adolescencije ili odraslog doba. Više od 40% pacijenata ispunjava kriterijume za poremećaj iz spektra autizma, poremećaj pažnje/hiperaktivnosti, ili oba. Takođe imaju povećan rizik od psihijatrijskih stanja kao što su anksioznost, depresija i šizofrenija. Pedijatri, kao lekari primarne zdravstvene zaštite, imaju ključnu ulogu u ranom prepoznavanju i dugoročnom praćenju. Preporuke za praćenje uključuju rutinske razvojne procene, rane razvojne intervencije (npr. govornu i radnu terapiju) i redovno procenjivanje koeficijenta inteligencije i adaptivnog funkcionisanja, naročito tokom prelaznih obrazovnih perioda. Rana dijagnoza i individualizovani, multidisciplinarni pristupi su od suštinskog značaja za unapređenje razvojnih ishoda i kvaliteta života dece i odraslih sa 22q11.2 DS.

**Ključne reči:** sindrom delecije 22q11.2, DiGeorge sindrom, neurorazvojni poremećaji

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