22q11 syndrome

Educational Handbook

Asociación Síndrome 22q11

COCEMFE

Confederación Española de Personas con Discapacidad Física y Orgánica

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ACKNOWLEDGEMENTS

We would first like to thank all the people with **22q11 syndrome**. Their motivation has pushed us to create this document, in which we attempt to provide an authentic description of who they are, how they learn, and what they feel. We wanted to share this knowledge with the entire educational community so that everyone with this syndrome can be seen, understood, and receive support at such an important stage of their lives.

A special thank you to each and every family whose children have **22q11 syndrome**. Their unconditional love and strength have inspired us to keep striving to make this genetic disorder visible.

A sincere thank you to Doctor **Sixto García-Miñaúr** (Department of Clinical Genetics, Institute of Medical and Molecular Genetics, INGEMM, Hospital Universitario La Paz, Madrid) for his availability and generosity in sharing his experience and vast knowledge of the **22q11 syndrome**, as well as his unconditional support to the **22q11 syndrome Association**.

We would also like to thank Doctor **David Fraguas** (Head of the Psychiatry Department at Hospital Clínico San Carlos, Madrid), for his thorough knowledge and empathy with people who suffer from **22q11 syndrome** and their families, for his timely availability and patience in answering our questions and, finally, for his assured consulting, which helped us during the preparation of this handbook.

We thank **Jaime Martínez de Pinillos López** (known as @jimilustra in social media), who brought our protagonists to light through his artistic illustrations.

As well as **Luis Miguel Yébenes**, father to a son with 22q11 deletion and headmaster of Santa María School, for his availability and contributions that have been of invaluable help.

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To **Nacho Liz**, for his valuable last-minute contributions and continuous support.

To all those who, either directly or indirectly, have made this handbook possible.

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PREFACE

As the mother of a child with **22q11 syndrome** and a professional educator, I have always been concerned that teachers may not understand my daughter or may tend to use unsuitable "labels". It is for this same reason that many families ask our Association to organise talks in educational centres, and that schools are increasingly frequently asking us for guidelines.

There is currently little information available in schools about this genetic disorder which is so frequent and yet so poorly understood. This is despite the fact that the best way to help students with 22q11 syndrome is to understand their situation in order to be able to empathize with them and thus promote their teaching-learning process.

This handbook is a tool for education professionals. We hope that it will enable you to understand the most common characteristics of students with **22q11 syndrome**, but also how different their profiles can be. It seeks to help you identify the manifestations of this disorder and provide you with methodological guidelines for working with these children and teenagers.

For many years, the Board of Directors of the **22q11 Syndrome Association** has been hoping to release this educational handbook in order to help families and professionals and, above all, the students themselves.

It has been a pleasure to be able to count on the help of great professionals who have supported, guided and advised us and who have collaborated with us repeatedly on various occasions. Their efforts and professional experience with people with **22q11 syndrome** were, simply, essential.

I would also like to mention here that the joint drafting of this handbook has proven of paramount importance to the authors. Without Lara's unconditional support and motivation, this handbook would not have been possible.

Children and teenagers with **22q11 syndrome** are no different from their peers. They have some specific characteristics and needs, and it is important for education professionals to know this, just like the rest of the students may have. Only when we understand this, inclusion can become a reality.

Our utmost thanks to COCEMFE for making this project possible.

Aldha Pozo,
President of the 22q11 Syndrome Association

FOREWORD

With approximately 7,000 genetic diseases and disorders considered to be rare, uncommon or of low prevalence, what distinguishes **22q11 deletion syndrome** (abbreviated to **22q11 syndrome**) from the rest? Based on my professional experience, broadly enriched over the past few years from the collaboration with a large group of specialists from abroad and our colleagues from the Institute of Psychiatry and Mental Health at Gregorio Marañon General University Hospital, I would focus on three key points.

- Its frequency, estimated between 1:2000-4000 births, which makes it the interstitial chromosomal abnormality of the highest incidence in humans.
- Its ability to go unnoticed, especially if it is not associated with congenital cardiovascular defects or palate defects.
- Its consistent (yet at the same time variable) pattern of learning difficulties and behavioural disorders, which allows close monitoring and anticipating problems, as well as an early and specific intervention.

During childhood, the school setting and interacting with peers are fundamental pillars of a child's development. Interventions aimed at improving social integration and academic performance of children with **22q11 deletion syndrome** will result, without a doubt, in a better long-term prognosis and a higher degree of personal autonomy as an adult.

Dr. Sixto García-Miñaúr
Clinical Genetic Unit, Institute of Medical and Molecular Genetics (INGEMM), Hospital Universitario La Paz, Madrid, Spain.

Poet Rainer María Rilke stated that the true country of man is childhood. Therefore both childhood and adolescence are essential because of our experiences and because they set the foundations of our adult life. But they are not easy. Far from rose-tinted (and always idealized) scenes from films and sentimental stories, they hide risky adventures and dangers that do not always end well and, frequently, result in emotional suffering. Children and adolescents with **22q11 syndrome** live these periods with much more difficulties than the majority of their peers. Almost everything is more complicated for them: They become ill and have to see a doctor, nurse or other healthcare professionals more frequently; it is more difficult for them to establish or maintain relationships with friends or classmates, and also to adapt to the changes in life; they face more learning problems and have difficulties entering the job market; they often show mental symptoms, such as anxiety and depression among others.

As always, family plays a key role in the emotional and vital warp and weft of people with **22q11 syndrome**. Family members, each one in their own way, support these children during their difficulties and share their joys.

This handbook constitutes an important step for all of them. It will contribute to improving their quality of life in school, a key element in their childhood and adolescence, thus improving their present and future.

Dr. David Fraguas
Head of the Psychiatry Department at the Hospital Clínico San Carlos, Madrid, Spain.

INTRODUCTION

«Every child has characteristics, interests, abilities and learning needs. Education systems should be designed and educational programmes implemented to take into account the wide diversity of these characteristics and needs.»
(Unesco, 1994).

The handbook you are holding in your hands aims at helping you get a better understanding of the people with 22q11.2 syndrome (hereinafter, **22q11 syndrome**), a complex genetic syndrome with neurodevelopmental disorders and great symptom variability. Like the majority of rare or uncommon diseases, it is quite unknown to the general population, despite the fact that its incidence is 1:2000-4000 births.

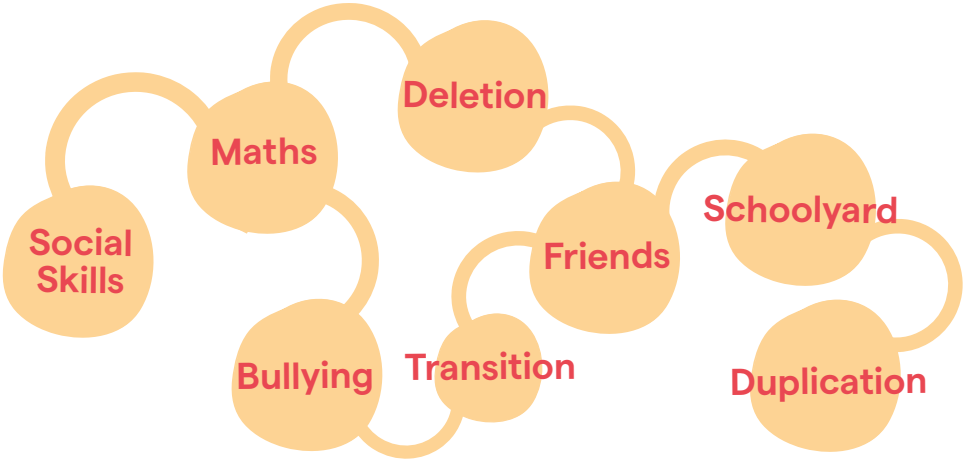
22q11 syndrome includes both disorders of loss (deletion) and gain (duplication) in chromosomal region 22q11, although normally it refers to deletions, which are associated with other congenital defects (heart, palate, etc.). While deletion has been known for many years, when the DiGeorge syndrome was described, identification of duplication only occurred recently, thanks to the development of new molecular techniques.

Its clinical manifestations are mild and variable. This handbook mainly refers to **22q11 syndrome** caused by deletion, with no intention to exclude the cases of duplication, for which limited data are available.

This work stems from the mission to offer a global vision of the main educational characteristics and learning needs you may see in students already diagnosed. It is essential that you have information on this syndrome and what it entails for your school, so you can spur their motivation to fully exploit their potential and reach their goals in terms of academic achievement and personal growth.

This knowledge, in addition to observing their individual skills, will help you avoid the use of wrong "labels" that conceal their actual potential.

The first part of this handbook introduces our protagonists, a group of students whose profiles have been outlined by the professionals who have collaborated with this project, summarising some of the more common characteristics of **22q11 syndrome**. However, we would like to highlight their great clinical variability and remind you that there could be as many profiles as people with this genetic disorder.



Afterwards we will show you some of the most relevant school situations that deserve special attention in order to help and understand these children. We have included some possible guidelines to intervene which could be useful in the classroom on a daily basis, for example, a description of strengths and weaknesses that the student with **22q11 syndrome** could show.

Lastly, if you would like to know more about this syndrome, please refer to the end of this handbook, which also delves into the differences between the disorders of deletion and duplication.

Let's get to know our protagonists.

1. STUDENTS WITH 22Q11 SYNDROME

Next, let us introduce you to Silvia, Leila, Javier, Ana... These schoolmates have been created by the professionals who have participated in the drafting and revision of this handbook based on their experience and data from studies consulted.

Each of these profiles will help you identify the most common situations children and teenagers with **22q11 syndrome** could come across daily. We will accompany them through the different stages of their school life, from childhood to adolescence.

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1.1 Profiles

Unai

Unai is five years old and he is in the third year of pre-school. His parents have visited various specialists since he was born and he has been recently diagnosed with **22q11 deletion syndrome.**

When he started pre-school, he still wore nappies and, currently, he does not control his urinary sphincter. At school they are planning to start working on getting him out of the day nappy in collaboration with the family. **Unai** suffers from a kidney problem, due to a malformation in his kidneys.

He also has speech delay. When he started school, at three years of age, he could say some single words.

Today he can produce some three-word sentences, but he is not able to speak in a fluent way as would correspond to the normal development of a child his age. He shows difficulty in both verbal and non-verbal communication.

He doesn't interact with his classmates, nor does he show interest in group activities.

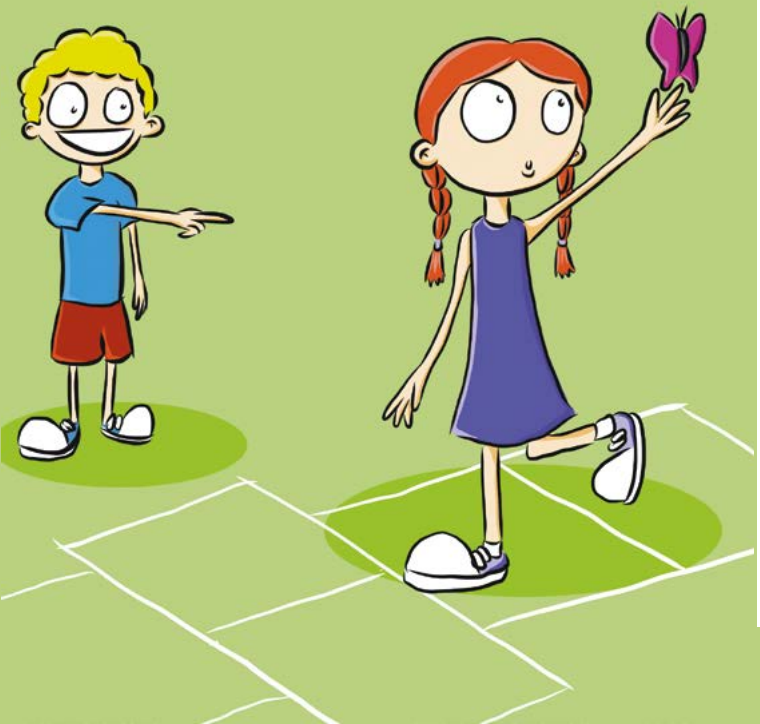
Unai, who is an only child, likes to play with wooden blocks; he could spend all day playing with them. He is a quiet child, but his teachers noticed that he sometimes gets agitated while sitting in his chair, without making noise, especially when he comes back from breaks or when there has been a special or different game during the break. He frequently becomes ill with fever and respiratory problems and misses a lot of school.

This year he started wearing glasses. He takes them off to play with the wooden blocks or when an activity involves effort.

In class, they are starting to read. He manages reading quite well, recognizing letters and starting to write his first words.

The orientation team of the centre, due to the difficulties he showed, spoke with his parents about it. They are very distressed because they have had many medical appointments since the diagnosis.

Silvia



Silvia is nine years old and she is in the third grade of Primary school. When she was two, she was diagnosed with **22q11 deletion syndrome.**

Since she left pre-school, she has difficulties concentrating on a task even for a few minutes. She gets distracted easily and finds it hard to resume an activity, because she didn't understand what she was supposed to do.

Her teacher says she can't stay still in her seat and she gets up many times to go to the bathroom or to take a walk.

It is difficult to understand her nasal voice. When she has to speak to a group or present a work, the tone of her voice is very low.

Silvia has scoliosis, a type of deviation of the spinal cord, and the doctors have notified her parents that in a few years she will have to wear a brace before thinking about surgery.

She is monitored by various specialists: among others, endocrinologist and nutritionist, due to her small size, calcium deficiency and thyroid problems, and she has always showed many "vagaries" when eating; traumatologist and rehabilitation specialist for her back; physical therapist; neurologist as well as immunologist.

She usually misses a few classes a month, although when she was younger, she had so many medical appointments that she used to miss even more. She gets very nervous when she is late for school and doesn't understand why she has to go to the doctor so often.

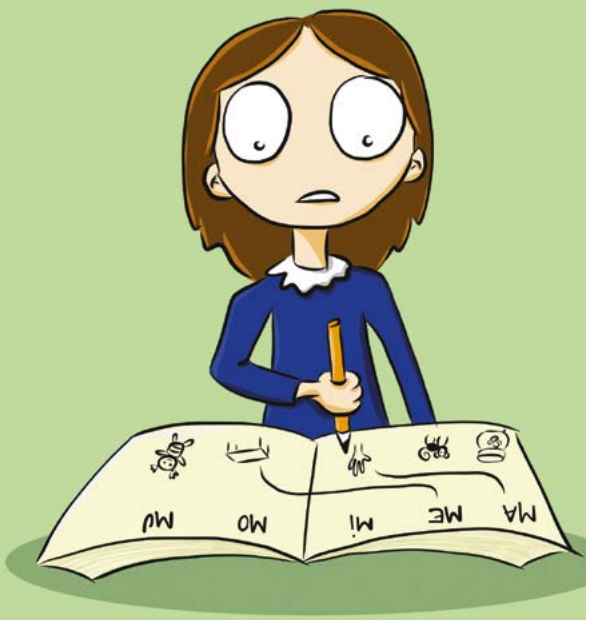
Due to the check-ups and therapy, she has never shared any after school activities or afternoons in the park with her schoolmates. When she is in the schoolyard or classroom and they make some kind of joke, she usually doesn't understand it. However, despite being uncomfortable, she stays with her classmates.

Nobody waits for her when going to the schoolyard and they don't really include her, but she seeks to be a part of a group. They have even made fun of her ears, which are small and oddly shaped.

Silvia has passed previous courses with non-significant curricular adaptation. This year she started noticing that she finds is very difficult, especially multiplication tables, which are impossible for her to memorize, and she doesn't understand the way problems are formu lated.

She likes Arts and Crafts and Music classes. She especially loves Music and her teachers say that she does quite well; she is highly motivated and pays attention and she has a good sense of rhythm.

Leila



Leila is eight years old and she is in the third grade of Primary school. When she was a month old, she was diagnosed with **22q11 deletion syndrome.**

She only started walking when she was two and a half years old. Nowadays she is very independent but not very agile in races or jumping. For this reason she dislikes Physical Education and it requires a lot of effort to follow the games her classmates are playing at the schoolyard.

She was born with a heart malformation that required surgery when she was five years old.

At the age of two, she also underwent surgery because she was born with a cleft palate. She still has trouble with the pronunciation of some phonemes and it is sometimes a little hard to understand what she says because of her high-pitched nasal voice. Leila attends speech therapy sessions twice a week outside school and receives the help of a specialist in language and hearing twice a week as well.

Leila was also born with hypotonia (poor muscle tone) and this affects her fine motor skills when writing, as well as her autonomy for doing things like button up her coat by herself etc. For this reason, adult supervision is needed. She has difficulty asking for help and on some occasions her classmates try to help her but she doesn't accept it. She would rather do things on her own and gets frustrated when she can't do something well.

After finishing first grade in Primary school her teachers and parents considered whether she should repeat the year since there were issues when she started reading and writing and she had missed so many classes throughout the year due to repeated episodes of otitis (she underwent auditory drainage with noticeable improvement).

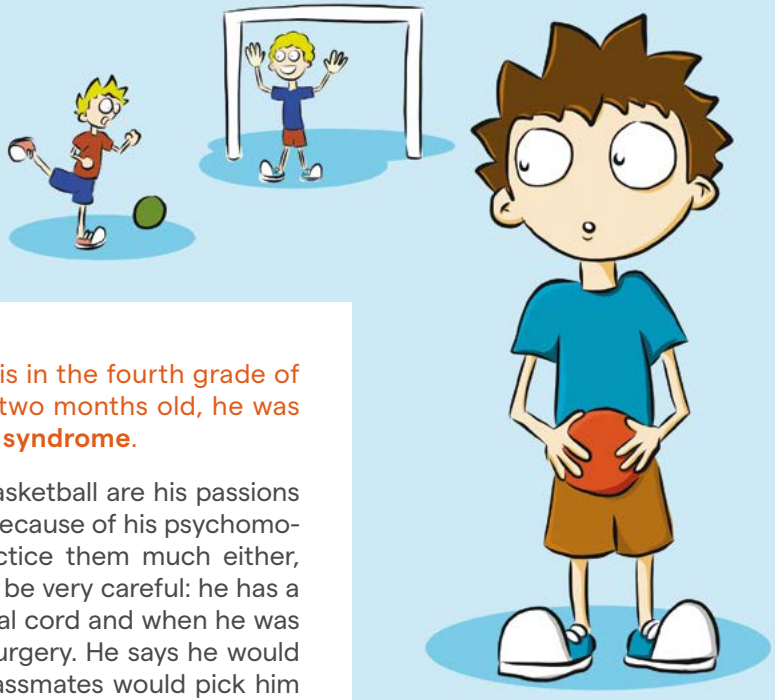
Leila shows dependence on a friend who is a year older, because her classmates don't play with her.

When this friend is not around, she looks for an adult.

She is not very flexible with change but she doesn't verbalise it. Sometimes when she arrives home, she says she has a stomach ache or a headache, and her parents notice that this happens when something has happened at school. The paediatrician mentioned she shows symptoms of anxiety.

Once a week she has a Music class. She loves it; she has a natural ability with rhythm and is learning how to play percussion instruments. Her music school teachers say that she is very affectionate with her classmates and educators and her parents have noticed that after these classes she is more relaxed and happy.

Javier



Javier is nine years old and he is in the fourth grade of Primary school. When he was two months old, he was diagnosed with **22q11 deletion syndrome**.

He loves sports. Football and basketball are his passions but he isn't very good at them because of his psychomotor difficulties. He doesn't practice them much either, because his parents told him to be very careful: he has a substantial curvature in his spinal cord and when he was younger, he underwent heart surgery. He says he would love to be better so that his classmates would pick him when they play matches and he could play with them during breaks, since he usually plays alone.

They don't make fun of him at school, but they don't integrate him into the group either. They never include him when making teams nor invite him to birthday parties. The situation has improved since the arrival of a social worker.

Javier would love to have friends to go to the park with and play on the playground. He doesn't know what is going on, but he thinks he is "dumb" or that they don't play with him because he has "a syndrome" (he overheard his parents and doctors talk about it, but he doesn't really know what it is).

Some days he tells his parents he doesn't want to go to school or do anything. His favourite subject is Social Sciences because he can easily memorize the cities and rivers in Spain.

His parents have signed him up for private classes, despite his keeping up with the rest of the class in the fourth grade; he is a year behind in Maths. However, since he started using an abacus, he is doing much better. Currently, he doesn't need any curricular adaptation.

He has had many epileptic seizures during his school years, in which he had to receive assistance by the school nurse.

Javier has had many medical checkups, which he doesn't like at all: on the days he goes to the hospital, he normally does not go to school.

Ivan

Ivan is twelve years old and he is in the sixth grade of Primary school. He was six when he was diagnosed with **22q11 deletion syndrome**.

Since the age of one, Ivan has been a restless boy. He finds it very difficult to finish any activity or game. Moreover, his parents say that he talks back to them at home. He was diagnosed with Attention Deficit Hyperactivity Disorder (ADHD) two years ago.

While in class, he makes comments that are out of line. The teachers don't know what to do, because he interrupts their explanations and alters the class rhythm. His classmates say he is very annoying and always disrupting class. This is why nobody wants to be with him in group or paired activities: Ivan gets angry and the teachers have to mediate to have someone be with him.

Ivan has a minor intellectual impairment. He has passed all previous courses with in-class support and non-significant curricular adaptation in various subjects, among which Maths and Language.

It is difficult for him to follow regular education and a decision must be made for the next year, when he will start secondary school: should he be forced to finish this stage, should a significant curricular adaptation be done in Primary school, although other alternatives will also be consided. The school centre has spoken to his family and they are assessing the best options for him.

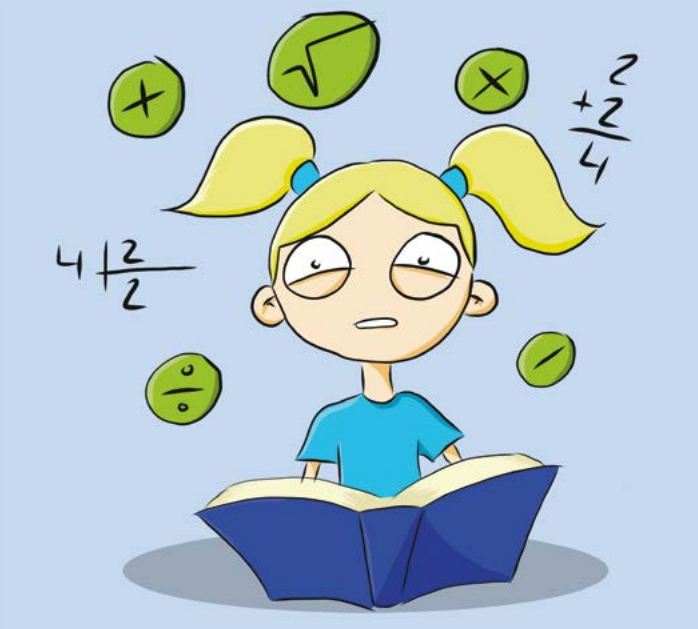
It seems like he enjoys being with his classmates even if he says he doesn't have any friends because he is not like them. The boys and girls have spoken to their teacher and told her that they find him annoying. They point out he doesn't understand the rules of the games and doesn't follow them.

He could be happily playing and the next moment he could suddenly get angry. This has provoked some arguments because they don't understand his mood swings.

At home he gets frustrated when he can't do his homework. He takes it out on his parents: shouting, throwing his pencil case around... Sometimes they can control the situation and he finishes his task; on other occasions they can't. In parent-teacher meetings his parents have said there is nothing more they can do in such situations.

Ivan would like to be a police officer or a footballer. He doesn't get on well with his ten-year-old brother (he seems to be jealous) and he doesn't want to play in the park because he doesn't have any friends to play with. He spends most afternoons with his cousins who he gets on well with and enjoys playing with them. He is very affectionate with them and they don't use to fight. He learns many things by watching them: he already knows the rules of football; how to make his bed and how to set the table.

Alicia



Alicia is a thirteen-year-old teenager and has just started her second year of Secondary school in a new institute. She was diagnosed with **22q11 deletion syndrome** when she was two years old.

Alicia completed Primary school with support but without any major difficulties. However, at the end of the course she started having problems with some subjects, especially Maths.

Her favourite subject is Language. Her teachers have told her she reads very well and she hardly makes any spelling mistakes.

Before moving on to Secondary school **Alicia** asked her parents if she was going to be with her classmates. That got their attention, but they didn't give it greater importance, so she did the first course in the same centre as in Primary school. However, once the course was over she said she didn't want to continue studying.

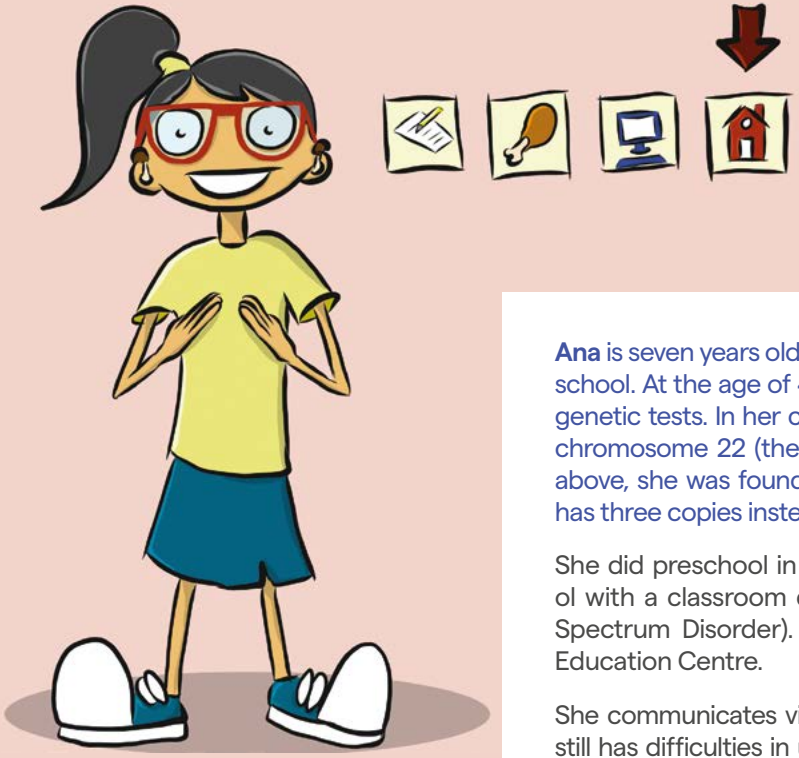
Her parents attributed it to the difficulties she was experiencing in her studies, and the fact she had a hard time passing each subject.

During that summer she said that she did not want to return to class with her classmates. This raised the alarm and her parents asked her if something had happened. **Alicia** told them that they were making fun of her way of speaking, her ears, the time it took her to finish exercises and because she wasn't good at sport. And on some occasions, they even pulled her ears. Given this situation they decided to move her to a smaller school.

During first grade of Secondary school she passed Maths with a lot of effort on her side and help from her parents. Alicia went to private classes and they dedicated many hours of the day helping her, especially with Maths, which she wasn't really good at. That's why she barely had any free time in the afternoons or weekends to share more playful moments with her classmates and she never managed to get integrated into a group of peers.

For **Alicia** this was "overwhelming", and "exhausting" for her parents, to the point of getting to "hate" Maths and developing symptoms of phobia.

Her parents are worried. They say it seems like she hasn't learned anything in Primary school, despite all the hard work and effort.



Ana

Ana is seven years old and she is in the second grade of Primary school. At the age of 4, a 22q11 duplication was detected in her genetic tests. In her case, instead of losing a small fragment of chromosome 22 (the 22q11.1 region), as in seen in the profiles above, she was found to have a duplication (that is to say, she has three copies instead of 2).

She did preschool in a regular school and Primary at a school with a classroom designed for children with ASD (Autistic Spectrum Disorder). Nowadays, she is enrolled in a Special Education Centre.

She communicates via pictograms and sign language but she still has difficulties in understanding orders and simple sentences (this is something they are working on in class and at home)

and she isn't able to have a conversation, although she has started to say some words ("mum", "school", "car"), which has improved her communication skills. Her parents are very happy because she throws less tantrums and she can explain and ask for some of the things she needs.

Ana uses a tablet in the classroom and thanks to ICT (Information and Communication Technologies) she is starting to improve at literacy. She can handles technological tools quite well.

Since she has hypotonia (poor muscle strength) she goes to the physical therapist after school. She also attends sessions with a speech therapist twice a week.

She needs help to carry out her daily activities and still hasn't achieved sphincter control; as a consequence, she needs support and attention from an adult all day.

She has been wearing a hearing aid since she was four, due to a sensorineural hearing loss. Moreover, she has been wearing glasses since the age of six, because she is far-sighted.

Ana doesn't usually become ill but needs medical follow-ups with her neurologist.

Alex

Alex is eight years old and he is in the second grade of Primary school, and like **Ana**, his genetic tests revealed **22q11 duplication**.

He doesn't meet the criteria for the diagnosis of an autistic spectrum disorder (ASD), but he does show some of its characteristics, such as difficulties in social interaction and communication and a limited flexibility.

He has communicative intention and communicates verbally. When he arrives at school, he talks about things at home, but sometimes his speech is incoherent. The teachers have observed that when he is asked closed, specific questions there isn't any problem; however, it is difficult for him to understand double meanings and he seems lost with open questions.

Every morning he does the same ritual when arriving to class. If not, he throws a tantrum, something that happens at home as well. If his mother cannot pick him up, as usual, the teachers have to tell him beforehand (as well as on many other occasions) in order for him to assimilate the information and organize himself because he cannot handle change well.

He learns very well through repetition and for this reason he stays for as long as possible in the classroom, and has assimilated classroom routines by imitating his classmates. He tends to get distracted and has difficulty concentrating, but when his teacher tells him what to do, he starts working without any problem.

Alex keeps up well with the rest of the class; he has a non-significant curricular adaptation to help him with the tenses and formulations, since he tends to make literal interpretations of questions and answers. He has difficulty with verbal and reading comprehension.

It is a very complicated for him to make decisions; this is why, both at home and at school, there is an effort to only have him choose between two options. However, it is still not easy - he gets very nervous and gets mental block.

He also has difficulties with social relationships. Almost always he plays alone on the playground, maybe because he has no interest in his classmates' games or because they do not integrate him in their group. He doesn't know how to do that either.

1.2 Manifestations of 22q11 syndrome through different profiles

With these eight children, our protagonists, we have tried to show you what they like and what they are good at, their fears and the common features of students with **22q11 syndrome.** They may have reminded you of a child whom you have worked with or have in your class, even if their profile is somewhat different. This depicts the great variability of this syndrome: each person presents specific features - and with different levels of intensity - as an individual and this is how they should be treated.

Now we are going to examine the features we have seen in **Alicia, Javier, Unai**... and briefly explain what they entail.

22q11 syndrome, as indicated in the introduction, could involve either deletion or duplication, which is less understood. Its clinical manifestations are mainly related to the different structures and organs involved, such as:

Congenital heart defects (like in **Leila's** and **Javier's** case, who underwent surgery at a very young age),

cleft palate or impaired palate function (resulting in **Leila's** operation and **Silvia's** nasal voice),

and **musculoskeletal disorders** (like **Silvia's** scoliosis or the deviation of the spinal cord Javier suffers).

They could be diagnosed with a **minor intellectual impairment**, like **Ivan**.

Some are prone to suffer from infections in the early years of life and the appearance of **autoimmune processes** make them feel sick frequently, as in **Unai's** case.

The majority attend regular education schools with educational support. They will need methodological and/or access adaptations mainly in Primary school without the need for significant curricular adaptations.

During the first years of life, they can show **motor skill delay**, with difficulties in acquiring strength and coordination, which is generally attributed to hypotonia. We saw this in **Leila**, who only started to walk when she was two.

When growing up, some continue having a **poor muscle tone**; this could have an impact on their motor skills, both gross motor skills (like Javier who isn't good at football) or fine motor skills (like Leila who can't put on her coat and finds it difficult to write, because she struggles to hold a pencil). This is why they attend early stimulation therapy, physiotherapy and occupational therapy sessions.

In **terms of speech development**, they usually don't say their first few words until they are two or even later, like **Unai**, who started at the age of three.

The majority show **delay in speech development** and **language problems**, for example, high pitched tone, hoarse or nasal voice and compensatory articulation errors. This is due to the problems some have with their palate. In **Silvia's** case, who has a very nasal voice, which often results unclear and unintelligible, or **Leila's**, who underwent palate surgery.

They are **literal thinkers and have difficulties** in understanding simple or implicit messages or phrases with double meaning, like jokes and irony. This happens to Silvia when her classmates tell jokes and she doesn't understand why they are laughing.

Attention problems are also common. It is difficult for them to understand relevant information and they get distracted easily. This is because they have a response inhibition deficit which affects their level of attention and manifests as distraction. This happens to **Silvia** and **Alex** in class: after a while they don't know what they have to do.

Children and teenagers with **22q11 syndrome** have difficulty in **solving problems** have difficulty in solving problems and applying the information obtained in new situations. They also have difficulty structuring information in a significant manner.

They can also encounter problems with abstract thinking while concrete thinking persists until more advanced ages, which affects performance in Maths and reading comprehension, like in **Alicia's** and **Ivan's** cases.

In Maths, problems are often bigger, especially for children who suffer from **22q11 deletion syndrome** (in the **22q11 duplication** they could have the same or minor impact), **spatial dyscalculia** is usually diagnosed, which appears at the age of eight or ten.

Memory is a strength for some of these children and teenagers. They are able to easily remember lists presented verbally and learning by repetition is also a strong point, just as is the case with **Alex**. However, this doesn't happen to everybody. In some cases, they cannot remember academic contents or other information worked on frequently, like **Alicia** whose parents complained that it seemed she hadn't learnt anything from the previous course, despite all her efforts.

In terms of **social interaction**, they might have little initiative with regards to social contact (**Alex**, for example) or an excessive familiarity, which provokes socially inappropriate situations (**Ivan**). Sometimes they have mood swings for no apparent reason: like **Ivan's** mood changes, which hindered relationships with his peers.

Due to the clinical manifestation of **22q11 syndrome**, medical consultations and different therapies may be frequent, especially since birth and up to the age of six years when they are most necessary. They even extend to the stage of Primary Education.

The average number of specialists the children see during their first years of life is around nine (cardiology, immunology, otorhinolaryngology, nutrition, genetics, etc.). Moreover, in some cases, different therapies may also be necessary (speech therapy, psychical therapy, stimulation, psychotherapy, occupational therapy, etc.) as well as surgical interventions.

It is important to empathise with this reality and understand how they may feel when they return to school. That is why communication between families and educators with respect to relevant medical information is fundamental. In the same way, educators must indicate the work they could not do during these absences, so that they can keep up with the pace of the rest of the group, when possible.

These students often are **emotionally dependent** on people or situations and are easily influenced because of this, like **Leila**.

Moreover, they often suffer from **motivation and self-confidence problems** due to academic difficulties and find it hard to socialise with their peers like **Javier** and **Alicia**.

Moreover, they usually lack the resources to face and defend themselves in hurtful social situations, which is why they are often victims of **bullying**, both physical and relational, because of their physical or intellectual differences, such as in the cases of **Alicia**, **Javier**, **Alex**, **Ivan** or **Silvia**.

In some cases, **perseverance of behaviour** and the lack of flexibility leads to repetitive attitudes or behaviour. This is due to the inability to modify their motor or verbal response or change the subject or activity, like **Alex**.

People with **22q11 syndrome** frequently suffer from **excessive anxiety** and worries which affect their working memory. They might show underlying anxiety, like **Leila** and **Unai**, without explicitly expressing it due to their verbal communication problems.

Children and teenagers with **22q11 syndrome** may need **social and health care** at school. The presence of a registered nurse is necessary in case it is required to administer medication prescribed by a medical specialist, or use of catheters, tube feeding, heart diseases, etc., as in **Javier's** case.

It is also important to have the presence of a **social inclusion worker**, to develop and work on social skills, personal autonomy and inclusion with peers, as in **Javier's** case.

NOTES

2. UNDERSTANDING STUDENTS WITH 22Q11 SYNDROME IN DIFFERENT CONTEXTS

2.1 Schoolyard

It is possible that for children and teenagers who suffer from **Syndrome 22q11**, time in the playground is not as useful and fun as for their classmates. In their case it might not be the atmosphere to allow them to interact with other children in a playful and relaxed manner, but instead it becomes a hostile place where:

- They are not able to form part of the group due to their difficulties in socialising, different social interests inherent to their age group and their immaturity that results in participating on unequal terms when interacting with peers.

- The games their classmates play do not always interest them and they seem "happier" by themselves. Attention must be paid to this in order to understand and differentiate their need to be alone sometimes from constant loneliness

- They can become victims of bullying with possible emotional repercussions (above all, anxiety or depression) and an impact on academic performance, which may result in their wanting to leave school.

- They might need more guidance from an adult, since it is a place with totally different rules from those in the classroom and they need someone to explain them so they can assimilate them.

- They might need an adult as a reference and guide so as to feel safe and encouraged when starting relationships with their peers.

For all these reasons, it is of utmost importance that educational centres implement a schoolyard project with the presence of a social integrator who will work on the diversity within the school setting and favour the inclusion of all students.

2.2 Bullying

Children and teenagers with **22q11 syndrome** are a group of people very vulnerable to bullying throughout their entire educational period. This risk may increase because of their difficulties in learning and socialising, physical features and their limitations in defending themselves and telling others what is happening. According to recent studies, 80% suffer bullying at some point during their educational period.

They can suffer two types of bullying:

Relational Bullying

Consists in not paying attention to or excluding them from social activities carried out at school (schoolyard, holidays, or special school days) or outside (birthday parties, extra-curricular activities, etc.). This usually entails medium to long-term negative emotional consequences.

Physical or Verbal Bullying

It is a more immediate and direct way of bullying. There are physical aggressions like pushing, hitting and stealing their school material, etc., or verbal aggressions like insults and humiliations. These situations are carried out in private or in public.

The repercussions of these two types of bullying on self-esteem are very deep and can be carried into their adult life.

It is very important for the educators to pay special attention to these students, since the majority don't directly verbalise situations of bullying they might be experiencing on a daily basis. Moreover, in their desire to be accepted, they might normalise them and not express them.

It is necessary to make the educational centres a safe place where children and teenagers can attend happily and peacefully. It should be a place where they feel respected and where nothing bad can happen to them.

2.3 Academic skills

As mentioned previously, the majority of students with **22q11 syndrome** attend regular education schools. It is very important to take into account their individual characteristics, needs and motivations in order to adjust the level of academic demand to the stipulated curricular goals.

Language

In this area, they have major difficulties learning from what they read (due to problems with reading comprehension), remembering facts, choosing relevant details and drawing conclusions. This is due to deficits in working memory and poor visuospatial ability.

Regarding writing, for some of them hypotonia can complicate tasks that require agility and meticulous control of movement, such as holding a pencil. They may also feel pain in their hands and wrists, which makes them feel more tired because they tend to apply too much force in the pencil grip.

In these cases, it could be effective to allow the use of tablets or let them write in capital letters for a longer period of time

Maths

Mathematics is usually the first area or subject in which they experience significant difficulties.

They often have difficulty performing visual and spatial tasks, as a consequence of deficits in working memory and impaired numerical processing functions.

They also have difficulties understanding and representing quantities, as well as accessing the numerical meaning of digits and symbolic (abstract) shapes. This is called spatial acalculia, it is due to a neurodevelopmental disorder and is characterized by deficits in the spatial representation of numerical information.

It is a neurological problem, since the evidence suggests that there are fronto-parietal network abnormalities, which means that it cannot be fully corrected by therapy, but rather it is a condition that will stay with them throughout their lives.

An approach to learning Mathematics from a visual, experimental and manipulative standpoint can improve their results. The use of abacuses or even computers and tablets can cover part of the difficulties caused by spatial acalculia, can motivate them and thus reinforce the learning process.

Other subjects

As for the rest of the curricular subjects, students with 22q11 syndrome can keep up with their peers, with support and non-significant adaptations, as occurs in Social Sciences and Natural Sciences.

In schools with a bilingual project, they may encounter difficulties, unless the second language coincides with their mother tongue. The English subject should be taught with the necessary support and / or adaptations, but the rest of the subjects should be provided in their working languages.

Regarding Physical Education, they may need some access adaptation due to their physical conditions. Sport time is a good opportunity to foster social relationships with peers.

These children and teenagers can follow Arts and Crafts and Music classes at the same pace as their peers, which can help to improve their self-esteem.

The rhythm and musical sense are some of their strengths. Thanks to its motivational ability, this subject allows them to work on social and emotional skills with their classmates, as long as the learning process in the classroom is carried out in an experiential way.

2.4 Educational transition: school centre, course or teaching method

Changing school, course or educational modality usually involves changes in their work dynamic, rhythm, timetables and so on, which requires a great effort of adaptation. That is why many **anxiety** symptoms may appear that hinder the perception of the school setting as a safe and welcoming environment and have a negative influence, especially in their motivation and performance.

It is important and advisable to maintain a stable group of classmates with whom the children and teenagers have had more affinity, when there are changes of course or stage. Keeping one or two friends is emotionally fundamental and will benefit their learning process.

These changes should be accompanied by preparation and involvement of the family and the school centre (both the educators from the previous course and those of the new one) and it is important to involve them in decision-making, offering information in advance and with clear explanations. The duration of these periods could be bigger in order to help them adapt correctly to the new environment.

2.5 Relationships with peers

Lack of social skills is common in children and teenagers with **22q11 syndrome**. They usually have difficulties in relationships with their peers. They may show interest in them but occasionally their low social competence hinders or makes it difficult to maintain them.

Their problems in context comprehension also complicate the situation. It is complex for them to assimilate and proceed to correct language comprehension; they don't understand jokes, irony, double meanings, some facial expressions or gestures.

On certain occasions their answers do not adapt to the situations and make for an erroneous judgement of the social interaction that is taking place and they make a comment that is out of line.

In order to favour their integration in the class and improve their learning and social skills, "great teamwork" is important (which will be done whenever necessary in a progressive manner, from small to larger groups) which also incites learning by imitation. To this end, whenever possible, necessary support will be provided inside the classroom.

It is important for educators to know the characteristics of students with **22q11 syndrome** when placing them in a group where they may fit in and in this way facilitate their inclusion.

It is important to promote the socio-emotional adaptation of these people and work on their social skills, which will allow them to interact with their environment in an adapted way. Imbalances are not as relevant in early childhood, but they become more evident as of the age of eight or nine.

It is advisable to pay special attention to discouragement or lack of interest in preadolescents, both in class activities and outside activities (excursions, group works...). Such lack of interest could be drawn upon the lack of significant social relationships or by the direct rejection from their peers. Some may even start to suffer from abulia (lack of motivation, energy and extreme indifference) for different motives, such as depression in their childhood and adolescence.

When changing to Secondary Education, it is necessary that the orientation team provide students and their families with information on educational itineraries available which can offer possibilities of continuation in their education

NOTES

3. WHAT CAN WE DO?

3.1 Some characteristics of students with 22q11 syndrome

- Borderline IQ or minor disability.
- Learning problems, especially in calculus and abstract reasoning.
- Problems with social cognition: difficulty to understand the social world and adapt to it.
- Problems with executive functions (planning, execution of complex activities, inhibition of impulses).
- Inflexibile way of thinking.
- Difficulties in handling emotions.

- Difficulty in controlling motricity.
- Increased risk of mental health symptoms, especially anxiety, depression, attention deficit hyperactivity disorder (ADHD), obsessive compulsive disorder, tics, autistic spectrum disorder features, psychosis.

22q11 deletion

22q11 duplication

- Learning problems, especially related to abstract reasoning
- Social cognition problems, although to a lesser extend as compared to deletion syndrome.
- Problems with executive functions (planning, execution of complex activities and inhibition of impulses), although to a lesser extend as compared to deletion syndrome.
- Inflexible way of thinking, with adaptation difficulties, although to a lesser extend as compared to deletion syndrome.
- Difficulties handling emotions. Difficulties controlling motricity.
- Increased risk of mental symptoms, especially anxiety, depression, attention deficit hyperactivity disorder (ADHD), and autistic spectrum disorder (ASD).

3.2 Strengths and weaknesses of students with 22q11 syndrome

Strengths

Verbal IQ
At school age, verbal ability is similar or better than that of their peers.

Language
Ability in expressive language.This aspect may be limited in case there are important difficul ties in pronunciation of the language.

Verbal and auditory perception
Good ability to recognize and dis-tinguish auditory stimuli as well as to recognize speech sounds.

Learning and memory through verbal repetition
Good results in verbal mechanical memory and mechanical learning. They have a good capacity to repeat after a period of time a list of components presented verbally.

Attention directed to only one point

Ability to focus easily on a single task without distractions.

Reading

Mastery of the early stages of reading and spelling.

Affectivity

They tend to have calm and affectionate attitude. However, some preteens and teenagers go through a period of irritability or behavioural disturbances that alternate with their usual affectionate attitude.

Sense of rhythm

Normally good development of musical talents.

Identification of writing

Ability to recognize letters easily and to start writing.

Imitation

Ability to imitate easily what they observe around them.

Computing skills

Good motivation and development of computer operational skills. Could be limited in cases of low or very low IQ.

Weaknesses

Intellectual Quotient (IQ)

Average IQ is 75 and close to 50% of children will be diagnosed with a minor intellectual impairment.

Verbal IQ

- Verbal memory: difficulty comprehending and handling more complex information, mainly involving long sentences, information sequences, orders, stories, etc.
- Language comprehension shows a minor or moderate delay of speech, with greater involvement of expressive language.
- The language used must be brief and specific and lack complex grammar, in spite of the probability of making actual grammar errors.
- Literal thinkers: they experience difficulties with subtle messages and implicit meanings (jokes, sarcasm and irony). They can understand literal phrases and expressions.

Non-verbal processing

In terms of non-verbal comprehension, they show difficulty in using and catching signs, like facial expressions, voice tone, posture, etc. to manage or qualify the information, which can affect social communication.

Hand-eye coordination

Difficulty in activities which involve precise controlled movements in which hands, fingers and eyes are used simultaneously.

Visuospatial organization skills

Difficulties in spatial orientation, object localization, understanding of spatial relations among objects or things (in front of, behind, right, left) and analysing the shape of objects.

Memory

In regard to working memory, they have difficulty maintaining information in their head while paying attention to something else.

Executive functionality and attention problems

- Difficulties in planning and organization.
- Sustained and selective attention: showing difficulty paying attention to relevant information and they are easily distracted.
- Response inhibition deficit which affects attention and appears as distractibility and difficulty paying attention.
- Distractibility: tendency to focus on minor details while overlooking important ones.
- Mental rigidity: deficit in executive functions inhibits the generation of alternatives and makes it difficult to deviate from the established order.

Difficulties in learning how to read

- Problems in reading comprehension.
- Difficulty in learning what they read, in finding it hard to understand, remembering facts, picking out relevant details and reaching conclusions. Could be due to memory deficits and impaired visuospatial skills.

Difficulties in voice and speech

- Most children have a deeper voice than their peers, as well as hypernasal speech because of the velopharyngeal dysfunction.
- Difficulty in the articulation of fricative or affricate consonants.

Learning difficulties related to Maths

- Difficulty solving visual and spatial tasks, working memory deficits, deterioration of numerical processing functions needed for the majority of Maths tasks.

  This is called spatial acalculia and it is characterized by deficits in spatial representation of numerical information.
- Problems in cognitive processing in the space-time domain, which includes basic activities like simple adding and subtracting.
- Difficulty understanding numerical magnitude, identifying or ignoring irrelevant information in the definition of a multiplication problem and multiply with precision numbers of more than one digit.

Poor muscle tone (hypotonía)

- Difficulty in rapid movements or reactions, which affects gross and fine motor skills.
- Writing: difficulty in performing tasks which require agility and a meticulous control of movements, like holding a pencil. Pain in the hands and wrists.
- Tired hands, due to excersing too much force when holding a pencil.

Difficulty in social interaction

- They may show emotional stability, but have difficulty handling their emotions.
- They have low tolerance to frustration.
- They are dependent on people and situations.
- They are easily influenced.
- They have trouble saying “no”: they have assertiveness deficit, related to emotional dependency, social cognition problems and difficulty constructing an identity.
- Difficulty learning social signs and rules.
- Difficulty in social communication:
- Problems in non-verbal communication, which involve difficulties using or catching signs like facial expressions, tone of voice, posture, etc., to direct and qualify they information they receive.
- Difficulty understanding jokes, irony and sarcasm.
- Extreme reactions in social interactions, from shyness to excessive familiarity.
- Low initiative in social contact.
- Difficulty socializing with peers.

- Difficulties noticing voice tone changes, significance, facial expressions and humour.
- Difficulties expressing mental states or identifying social situations.
- Risk of internalising emotional problems (related to the possibility of somatising, developing phobic symptoms, sadness, mood swings, obsessions, etc.).

Behaviour

- The perseverance in their behaviour involves repetition of an attitude or behaviour due to the inability to modify their motor or verbal response, or change the subject or activity.
- Difficulty self-regulating and handling emotions.
- Changes in routine: they feel uncomfortable when facing new situations, as a result they experience anxiety and distress.
- Difficulties when asking for help.
- They suffer from extreme anxiety and worriedness, which affects working memory. May show underlying anxiety; not expressing it explicitly due to oral communication problems.

3.3 Methodological guidelines for teachers

Cognitive development

- Reinforce direct learning by repetition.
- Structure the tasks in short periods of time, wait for them to finish before giving them a new task.
- Promote visual contact with the adult and make them feel close to the teacher.
- Always give them free time when possible so that they could take a break and move freely.

- Provide a structured and predictable environment.
- Anticipate, inform and plan with each student the structure and content of each activity, indicating when they will start and finish it.
- Pay attention to signs of underlying anxiety.
- The teacher should practice the capability to disable their impulses through external regulation.
- Help them be constant in fulfilling their responsibilities.
- Amplify their centres of interests to avoid behavioural rigidity.

Behavioural development

- Place them in the front rows, but with full view of the classroom.
- Use direct and short commands.
- Breakdown the formulations and parts of any exercises.
- Vocally remind the commands and make sure they understand them (we could ask them what they have to do, or what we have just spoken about, etc.).
- Pay attention to their effort, try to do the more challenging tasks at the beginning of the day.
- Provide structured environments.

Academic skills

Academic skills

- Anticipate changes whenever possible.
- Use visual material that can be manipulated.
- Apply experiential methodologies whenever possible.
- Apply repetition methodologies.
- Promote the use of information and communication technologies.
- Adapt the classroom or materials in case of motor impairment difficulties.
- Use thicker writing and painting materials if necessary.
- Allow support from an adult as long as it is necessary to initiate the task or stay close during its performance.
- Praise achievements and motivate the students.
- Make them feel confident and feel that they can do it (no matter at what pace or in which way).

Academic skills

- Interaction of the different educators so as to adjust the quantity of homework to the capacity of each student.
- Respect their pace when writing in their diary or taking out books and notepads, performing activities, etc.
- During exams you should:
  - Ask clear unambiguous questions.
  - Read out the questions and ensure they have understood them (by asking them what they have to do). If they haven't, repeat or explain.
  - Highlight the key words in the formulation of the questions.
  - Present activities using images.
  - Ask questions which can be answered in few words, since they have difficulty sequencing ideas.
- Involve the family so they can work the texts at home and talk about the aspects they will discuss in class.
- Whenever possible, always evaluate orally based on the work done both in the classroom and at home, either individually or in small groups, and complement questions with pictures, etc.

Academic skills

- Maths:
  - Provide templates with the steps to be taken to solve the problems.
  - Limit operations shown in boxes.
  - Use the abacus or other visual kinaesthetic methodologies.
  - Allow the use of a calculator.
  - Facilitate learning by repetition of mathematical algorithms, how and when to apply formulas.
  - If they know how to apply formulas, but have forgotten them, allow them to have access to them during the evaluation.
  - Provide a clear verbal description thus substituting the intuitive, spatial or relational comprehension skill they lack.
  - Use of technology.

Academic skills

- Reading:
  - Promote learning in small groups or individually.
  - Practice with sensory support.
  - Provide information about what they are going to read (talk about the topic, watch a video, etc.).
  - Facilitate the use of new technologies.
- Writing:
  - Avoid homework that involves only writing.
  - Allow and facilitate that they can present or perform their activities on a computer.
  - Delimit the spaces for answering taking into account their dysgraphia.
  - Allow writing using a keyboard when needed.

- When necessary, support the language with augmentative and alternative communication systems (AACS).
- Sign language can be used at the beginning of language acquisition.
- Support oral language with images.
- Avoid fixed phrases or idioms.

Language and speech development

Social skills

- If necessary, promote time in the schoolyard being accompanied by educators or referenced colleagues.
- Make breaks meaningful, offer alternative activities.
- Establish clear and structured classroom rules.
- Provide models of adequate emotional responses (learning by imitation).
- Create a peaceful atmosphere in order to avoid tensions.
- Try to maintain the same close classmates in the same class when changing courses.
- Work on specific situations, let them know in real time when someone is joking or how someone is feeling in certain situations.

- Let the families know what situations and social difficulties the student experiences, in order to work on them at home.
- Develop conversational skills activities (describing, talking about current topics, etc.).
- Practice role-playing as a group dynamic, which allows working on empathy and social skills. Show them how to identify other people's interests in order to speak with them; offer them questions to facilitate their initiating a conversation.

Social skills

NOTES

4. WANT TO LEARN MORE ABOUT 22Q11 SYNDROME?

4.1 22q11 deletion

22q11 deletion is a submicroscopic chromosomal abnormality of a high incidence, with an estimated frequency in the general population of 1:2000-4000 newborns, although it might be higher.

"Deletion" refers to the loss of a fragment of chromosomal material and of the genetic information it contains (coming from the Greek word "deleterious" meaning to "destroy, eliminate"), in this case in the region of chromosome 22q11. The reason behind the great variability of the clinical manifestations of this disorder among people who have lost the same quantity of genetic material is still unknown. It is thought that there might be other, yet unknown, genetic factors which modify the impact of deletion on each individual. It is expected that more information will become available over the coming years.

The manifestation of **22q11 syndrome** is mainly related to the different structures and organs involved:

- Congenital cardiac defects (50-75%).
- Cleft palate or velopharyngeal insufficiency (60%).
- Calcium regulation disorders with a tendency towards hypocalcaemia or a drop in blood calcium levels (50%).
- Susceptibility to infections during the first years of life and to the development of auto-immune diseases.

The presence or absence of these problems and their seriousness varies from one person to another. Moreover, there is no correlation between them, meaning that the existence of a cardiac defect does not entail a higher probability of suffering from palate problems or a delay in psychomotor development.

Apart from such medical issues, which are rather more relevant during the first years of life, people with **22q11 syndrome** show some special characteristics in their psychomotor development and quite specific learning difficulties, which have been described throughout this handbook.

A delay in language development is very common (80%), regardless of the existence of a palate-related problem or not.

During school years a deficit in working memory is frequently manifested, which makes it difficult to process information and hinders abstract reasoning necessary for problem solving, reading comprehension or mathematical calculations. Despite the fact that such deficits are very specific, they may go unnoticed by teaching staff.

Behavioural disorders such as attention deficit, anxiety, oppositional defiant disorder and obsessive-compulsive disorder are also frequent. All these make students more vulnerable at school, especially in periods of greater social and academic demand, such as adolescence.

Learning and social integration problems cause a great deal of concern within their families. 22q11 deletion predisposes the appearance of psychiatric disorders in adult life as well. The frequency of schizophrenia in adults with **22q11 syndrome** is 20 times higher than in general population.

4.2 22q11 duplication

Duplication is a chromosomal abnormality complementary to deletion and is produced by the same molecular mechanism. It is the other side of the coin. Its symptoms are usually minor and less recognisable, which may cause it to go unnoticed. Duplication is far more difficult to detect using the FISH technique, which has been for many years the typical way to diagnose 22q11 deletion. The development of new molecular techniques like MLPA or array-CGH have made its identification possible.

Unlike deletion, whose diagnosis can be suspected in the majority of cases because of the pattern of medical problems and certain facial features, duplication is identified in patients studied for different reasons. Such reasons are also associated with deletion but they can be unspecific when considered individually (small size, learning difficulties, congenital cardiac defects, autistic spectrum disorder etc.).

22q11 duplication is currently considered a genomic variant susceptible to neurodevelopmental disorders, often inherited from an asymptomatic parent.

NOTES

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Area manager, graduated in Social Work, master's degree in Social Services and Welfare Management. He holds different qualifications related to team management, leadership and labor mediation. He has more than 30 years of experience in management and execution of social projects in different communities, more than 25 years in team management and more than 20 years in creation and development of projects for the disabled.

Rocío Bordoy Lobo

Education Programme coordinator, she holds a degree in Social Education, a master's in Psychomotricity, a master's in Early Attention, she is an expert in Therapeutic Psychomotricity, expert in Employment Guidance and master's in Dance Movement Therapy. She has more than 20 years of experience in social intervention projects in vulnerability settings and in cases of social exclusion with people with or without disability in different institutions and more than 5 years of experience in therapeutic body intervention in different communities focused, among others, on inclusion.

Cristina García Núñez

Technological tutor of the Education Program, holds a degree in Education, graduated in Psychology, Education Psychology, Health Psychology and Intervention in Mental and Behavioural Disorders. She has received various trainings about students with specific needs and learning difficulties. She has more than 9 years of teaching experience working on inclusion of students and fostering the use of innovative methodologies which promote integrated development from early childhood.

Maryorie Benavides Benavides

Technological tutor of the Education Program, holds a degree in Preschool Education and she is an expert in ICTs adapted to special education and in music therapy. A great part of her experience has been focused on working to improve the quality of life for those physically, organically or intellectually challenged. She has a wide experience in educational technology projects, in which she has collaborated with different Spanish entities. More than 9 years of experience working for the full inclusion of the disabled and promoting inclusive methodologies in the classroom.

22q11 Association Education Team

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Aldha Pozo Rodríguez

President of the 22q11 Syndrome Association and a mother to a girl diagnosed with 22q11 syndrome, she holds a degree in Hearing, Language and sign language interpretation, and has studied Psychopedagogy and Social Integration. She has an extensive experience in intervention and management in the field of disability, both in public and private institutions. She has more than 10 years of experience in the development of educational projects in regular education centres as well as Special Education centres related to the field of hearing disability.

Lara García Cela

Responsible for the Information and Educational Guidance Service of the 22q11 Association, she holds a degree in Pedagogy specializing in Special Education, she is expert in Early Attention, has been awarded a master's degree in Management of Social Services Centers and a masters in Music Therapy, and has studied Psychology. She has different qualifications in specific needs for learning and educational innovation. She has extensive professional experience in training and education management and has more than 10 years of experience in intervention in different communities mainly focused on people with disability and their families, developing and implementing family workshops.

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Technical collaboration and revision

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David Fraguas

A psychiatrist, head of the psychiatric department of the Institute of Psychiatry and Mental Health at the San Carlos Clinical Hospital in Madrid and coordinator of the mental health services in the Central District of Madrid. He is also a researcher at the Mental Health Networking Biomedical Research Centre (CIBERSAM) and an associated professor in the School of Medicine at the Complutense University in Madrid. David Fraguas has experience in the clinical assistance of people with neurodevelopment disorders and with genetic syndromes like 22q11 syndrome. He participates in national and international research projects dedicated to mental health of people with 22q11 syndrome.

NOTES

For more information and resources, please visit the 22q11 Syndrome Association website:

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