# PARENTS' GUIDE

THEORY TEACHES, EXPERIENCE TELLS

# SLEEP

in children with Williams Syndrome
DIALOGUE BETWEEN THE KNOWLEDGE OF OPERATORS AND THE EXPERIENCE OF PARENTS

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IN COLLABORATION WITH

Associazione Persone
Williams Italia

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# PREMISE

The professionals of the Davide e Golia association have been collaborating for years with Williams families and the Williams Syndrome Association of Italy, carrying out observation, rehabilitation, and monitoring projects for children and young people.

This collaboration has allowed the developmental neuropsychomotor therapists working at Davide & Golia to meet numerous families of children who came to us at a very young age and were then supported on their journey to adulthood.

Over the years, observing and caring for over 40 children has allowed us to gain a solid understanding of Williams Syndrome, enabling neuropsychomotor therapists and other staff professionals to broaden, deepen, and update their knowledge and share it with families and educational and rehabilitation professionals.

From the request to disseminate this specific knowledge in seminars and conferences, the need emerged to give structure to the experiences and experiences integrated into scientific knowledge.

In response to families' requests to leave a record, this project was born, aiming to capture and gather the preciousness of this knowledge with the intention of sharing it in the form of advice and suggestions.

The book also includes the results of a survey conducted among families specifically on the topic of sleep, allowing suggestions to be made based on the concrete experiences gathered.

# WHY DO WE TALK ABOUT SLEEP?

Over the years, we have realized that the stories regarding falling asleep and sleeping patterns of children with Williams syndrome are similar, as are the attempts made to manage it.
Sleep is a fundamental part of infant and child development, and gathering information on this aspect always deserves dedicated time for all the children we observe.

Let's start by talking about sleep because of the significant impact that good rest has or can have on daily activities, and especially for our children, including on the learning of new skills and on their development in general.
Equally important is good parental rest: a series of sleepless nights leads to irritability and a lack of availability toward the child, who in turn becomes more irritable and difficult for the tired parent to manage. Therefore, the first strategy we suggest, where possible, is to rely on grandparents, aunts, uncles, or friends who can help parents, allowing them to recover and better cope with daily life.

# THEORY TEACHES...

Before talking about "good sleep," we feel it's necessary to make a brief theoretical reference to "sleep function," understood as a complex function involving various parts of the central nervous system, the subject of numerous research and studies.
The sleep-wake rhythm is in fact regulated by the activation of various brain areas.
Various stages of sleep can be distinguished in the transition from wakefulness to sleep, and it is essential that each stage is experienced in its entirety before moving on to the next stage.

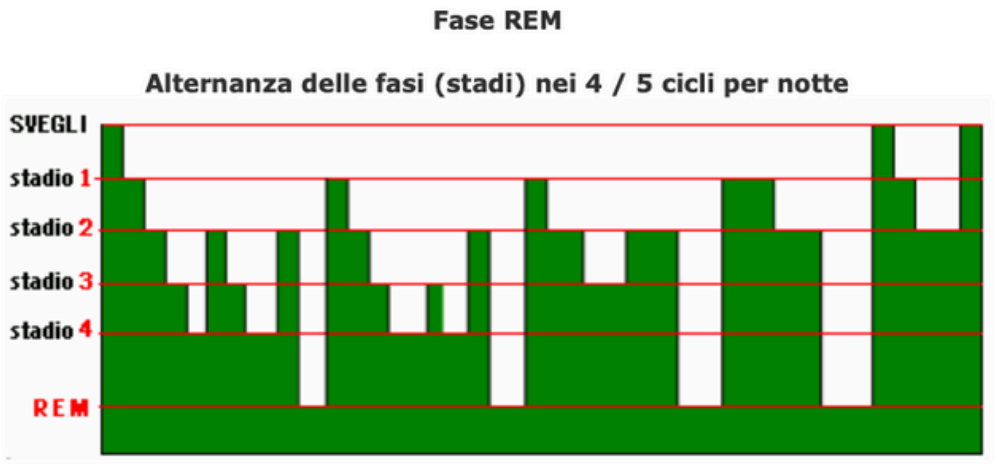
From the analysis of electroencephalograms, we know that each stage involves different electrical activity in the brain. Basically, two main phases are recognized: non-REM sleep and REM sleep.

There are four distinct stages of non-REM sleep, from falling asleep, through light sleep, deep sleep, and finally deep sleep. This is the stage in which our body truly regenerates and is the most restful.

During REM sleep, the eyes move in rapid, rhythmic movements. This phase, which typically occurs 4 or 5 times a night, is characterized by very intense dreams. Sleep during this phase is called paradoxical sleep because of the high brain activity and rapid eye movements, which contrast with the general degree of muscle relaxation.

During the night, several sleep cycles occur, lasting 90-100 minutes, characterized by the transition through various sleep stages and the REM sleep phase. Over the course of the night, deep sleep phases progressively decrease, while REM sleep phases increase in duration and intensity.
The first period of REM sleep is the shortest (5-10 minutes), while the last one can last up to 40 minutes (the one closest to waking up).

Various sleep studies agree that both REM and non-REM sleep are essential for good health, but the specific role of each is still unclear. We know that during non-REM sleep, there is increased production of growth hormone, which is vital for physical health, while REM sleep increases blood flow to the brain, which is beneficial for mental health. If a person is disturbed during REM or deep sleep, they are likely to experience symptoms of stress or nervousness.




A baby's sleep-wake rhythm in the first months of life is very different from that of an adult; in the first few months, babies don't know the difference between day and night: their rhythm is independent of the environment, regulated by internal needs related to hunger and thirst, and lasts around 25 hours. Newborns need to eat frequently, with a hunger-regulated rhythm lasting about 3-4 hours, regardless of the time of day or night. Their frequent awakenings are modulated by the sense of hunger and satiety, but also appear to be genetically determined.

The length and structure of sleep cycles varies with age, and a child's sleep cycle is much shorter than an adult's: in the first few months, babies experience a greater amount of light sleep than adults.

In newborns, a sleep cycle consists of an initial portion of REM sleep and a second portion of non-REM sleep. In the first two months, a sleep cycle lasts approximately 50 minutes and is evenly distributed throughout the 24 hours. A succession of 3-4 sleep cycles allows for a 3-4 hour sleep period. 18-20 sleep cycles occur over the course of a 24-hour period, with no daytime or nighttime pattern.

At 6 months, cycles last 70 minutes and are mainly concentrated in the night hours; at this age, non-REM sleep already begins to differentiate into a lighter phase and a deeper phase.

Gradually, the cycles lengthen until they reach 90-120 minutes and occur approximately 4-5 times per night during a normal adult sleep period of about 8 hours. Newborns sleep approximately 16-18 hours a day, and sleep is evenly distributed throughout the 24-hour period. However, there are significant individual differences: some children sleep 20 hours, while others need 14 hours; some begin to sleep continuously throughout the night, while others wake up every 30-60 minutes.

If we evaluate the development of sleep we see that the most important changes occur in the first 6 months, subsequently the variations are minimal:

- - Between 1 and 6 months, a day-night periodicity appears, wakefulness is distributed in the late afternoon and evening, progressive modifications are determined in the circadian rhythms of temperature, cardiac and respiratory activity and hormonal secretions; sleep matures on the electroencephalographic level and is structured in a similar way to that of adults.
- Between 4 and 6 months, a baby can begin to sleep up to 6 hours continuously at night, can stay awake longer during the day, and begins to be influenced by the light-dark rhythm. The total amount of sleep is 12-14 hours, distributed mainly during the night hours.

- Between 6 months and 4 years, sleep time progressively decreases to 10-12 hours, including nighttime and daytime sleep, and wakefulness increases. At 1 year, the child sleeps 13 hours, and between 3 and 4 years, 12 hours. The number of naps decreases from 3-4 at 6 months, to 2 at 12 months, and then to just one afternoon nap, a slightly longer one, at 18 months.
- Around 9 months, there is an increase in night wakings between 9pm and midnight and between 3am and 6am, which often continues until 2-3 years of age.
- Between the ages of 5 and 12, children experience the "best" sleep and the greatest capacity for alertness and attention. Sleep evolves toward an adult pattern lasting between 8 and 9.5 hours. The sleep structure is more stable, and daytime sleep disappears (note 1).

In addition to delays in various developmental milestones, children with Williams syndrome may also exhibit atypical sleep-wake rhythms. From information gathered through a questionnaire administered to families (details of the responses are attached), it can be deduced that sleep disturbances are reported during the first years of life.

Sleep disorders, as classified by the DSM V2, include 10 disorders or groups of disorders, for the diagnosis of which the presence of multiple symptoms is required.

In our cases, it's rare for all diagnostic criteria to be met, so a true diagnosis of a sleep disorder is rarely made. More often, the symptoms are difficulty falling asleep, staying asleep, or frequent awakenings.

It should also be said that Williams Syndrome is often diagnosed after the child's second birthday, so parents often find themselves dealing with the challenges of a sleepless child, but who hasn't yet had any diagnostic doubts or been tested for larger, more specific issues.

Note 1 - Booklet edited by Dr. Oliviero Bruni, Center for the Study of Sleep Disorders in Developmental Age, University of Rome "La Sapienza"

Note 2 - Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition, 2 of the American Psychiatic Association

In Williams Syndrome, various factors may be present that can affect the regulation of the sleep-wake rhythm, or more simply the ability to rest well.

Our babies often suffer from reflux problems, or even simple infant colic, which hinders sleep and causes frequent awakenings. Another aspect worth considering is hyperneurosensory sensitivity. Hyperneurosensory sensitivity refers to a condition in which a person has a high sensitivity threshold across the five sensory channels.

Several studies have been published on this topic, aiming to investigate the anatomical correlations with the various disorders found in individuals with Williams Syndrome.

One of the main symptoms that fall into this sphere is hyperacusis, understood as a hypersensitivity of hearing abilities, in the presence of a normal hearing threshold.

Several studies have been conducted to identify the main causes of this characteristic. Most authors favor an alteration in the integration processes at the level of the central nervous system, which also affects other cognitive abilities. Supporting this hypothesis are also some data on the correlation of brain area activation studied using functional magnetic resonance imaging during auditory stimulation with music and noise.

Also particularly important are studies on the levels of NGF (Nerve Growth Factor), which are found to be high in the serum of patients with Williams Syndrome even in school-age and adolescence.

Neural growth factor (NGF) regulates the development and survival of neurons. During the pre- and postnatal period, the normal development of peripheral sympathetic neurons and sensory neurons depends on the availability of NGF, produced by the target organ.

It would also be responsible for cell growth, neuronal production and elongation, and selective cell death and survival. Maintaining high levels of NGF during development would cause hyperinnervation of target organs, various neural dysfunctions, and others (Note 3).

In a 2000 study (Calamandrei, Alleva, Cirulli, Queryas, Volterra, Capirci, Vicari, Giannotti, Turrini, and Aloe), some phenotypic and clinical characteristics are described in light of the results of neurobiological analyses. In particular, with regard to NGF, levels were significantly higher in individuals with Williams Syndrome compared to control groups. Specifically, unlike controls, whose NGF levels were high in early childhood and tended to decrease with age, individuals with Williams Syndrome maintained high levels of the neurotrophin from early childhood until adolescence. According to the authors, hyperacusis could be secondary to the altered level of circulating NGF, as this would result in "hyperinnervation" in the inner ear (Note 4).

Their extreme sensitivity to touch, especially in their toes and fingers, and their frequent hypertension, could also be interpreted on the basis of the high level of NGF.

This extreme sensitivity can often lead to poor tolerance to contact with surfaces of varying textures, temperatures, or consistencies. In everyday life, this translates into difficulty having one's nails cut, as well as difficulty walking barefoot on different surfaces, such as warm beach sand or a cold floor. Likewise, it is this extreme sensitivity that causes difficulty or discomfort in wearing clothing with labels in contact with the skin.

Returning to the subject of sleep, if the hyperneurosensorial nature described so far can be defined as a constantly on switch, we can understand how much this can influence the ability to fall asleep and then maintain it for an adequate amount of time to allow for rest.

Notes 3 and 4 - Williams Syndrome. Clinical, Genetic, and Rehabilitation. Edited by Giannotti, Vicari, and Franco Angeli, 2004

# EXPERIENCE TELLS...

Starting from this theoretical basis, let's see what are the most frequent situations that parents of a child with Williams Syndrome find themselves facing in the first years of life.

An analysis of the questionnaire responses reveals that the greatest difficulties are encountered in falling asleep and maintaining sleep, which is interrupted by frequent awakenings. In the free narrative, parents sometimes report that sleep is good because, when adding up the various naps, the amount of sleep seems adequate.

In reality, as seen above from the description of the entire sleep cycle, we know that frequent awakenings interrupt sleep, preventing the child from reaching the deep sleep necessary for rest. At the same time, frequent awakenings do not help children regulate their sleep-wake rhythm: it is important that all the various sleep cycles (described above) are achieved and follow one another (non-REM sleep, REM sleep, etc.) for sleep to establish a regularity that, in the long run, leads to a complete and satisfying rhythm for a good quality of life.

When discussing sleep issues, families often report that they have "tried everything," describing all the various strategies they have implemented to help their child sleep.

Our role becomes that of trying to give meaning to the child's various behaviors in order to guide parents in finding the best solution for the specific need of that moment.

Generally, the first thing to discuss concerns the quality of the environment and preparation for sleep, both in terms of the space in which to sleep and the time spent getting ready to sleep.

For every child, the environmental characteristics that facilitate sleep are different, and only by carefully listening to the child's parents' stories can we suggest ways to try them, because trial and error is also necessary.

In doing this, as well as in seeking a meaning to the child's behavior, we must always refer to the specific theoretical knowledge of Williams Syndrome.

Let's take some practical examples.

Starting with time, the first step is to create routines, defined as a sequence of actions always performed in the same way and in the same order. This allows children to recognize, predict, and therefore prepare for what is happening.

In addition to encouraging routine activities, it is important to prepare the environment and the child by reducing activating suggestions and selecting stimuli at a specific time before falling asleep.

In fact, it is necessary to limit the intensity of stimuli (sound, visual, tactile, proprioceptive, etc.) to create the optimal conditions in which the child can fall asleep.

We can safely say that the rituality of the sequence of actions and the way they are presented makes all the difference compared to anything that is presented in a discontinuous manner. This applies both to nighttime sleep and to afternoon sleep, when it becomes a single nap.

Some parents talk about the importance of creating a routine or ritual that includes bathing, feeding, and bedtime, and how this makes it easier for the child to fall asleep.

For some, massage after bathing or before bed was also important, as a moment of relaxation but also a way to connect and share pleasant moments with their baby.

During this time before falling asleep, it is also important to pay attention to the confusion that can arise from the crowding of voices, sounds, lights, images and movement (the television on with the little brother running and screaming, the dog barking, the coming and going of several people...).

In some cases, it was important to select pleasant or inviting personalized music or rhythms, combined or not with soft or intermittent lights.

In others, using particular noises (white noises, such as a hair dryer, vacuum cleaner, etc.) or adjusting the lighting (lights with different intensities and movements, darkening the room, etc.) may work.

Particular attention must also be paid to the information a child receives through touch. We know that touch is one of our children's ever-open channels. Therefore, the characteristics of the surfaces the child comes into contact with become important.

Skin-to-skin contact with the parent certainly brings reassurance, in addition to the physiological exchanges that this allows (we will not address here the meanings given to the importance of physical contact between mother and child from the very first hours of life).

One of the main characteristics found is the need for physical contact: this leads to the need for a parent to always be close to the child to help him sleep, which then translates into organizing shifts during the night, with the loss of sleep for one of the parents.

Another containment method often reported in questionnaires is rocking, understood as holding (holding the baby in your arms) but also rocking him in different ways: bouncer, pram, stroller, even trying different speeds and movements.

However, there are also some precautions in preparing the cot/crib space, which can facilitate falling asleep.

Generally, in the experience of our parents and children, all those measures that concern the limitation of the space where the child sleeps, for example the boundary delimited by pillows or rolls, but also the consistency of the mattress, the fabric of the sheet, etc., have contributed to this goal.

For example, in one situation, it worked by putting the baby to sleep directly in your arms, but on a pillow, which was then placed in the crib with him. This avoided changes in contact and temperature, allowing the environment to remain constant and thus prevent sleep loss.

# CONCLUSIONS

Even if a solution isn't found, we believe it's crucial to support the family during this time, which we know is temporary but extremely difficult, and to avoid making them feel alone and helpless in their apparent inadequacy.

Experience tells us that sleep difficulties are real and present, and therefore significant, but they don't last forever. They certainly impact the first years of life, and impact the child's life, but also the daily life of the entire family, requiring a general reorganization.

We know that these issues eventually pass, but we also know that it's necessary to pay due attention to this aspect and try to find solutions, measures, or adaptations that are effective and true for each individual child.

Understanding the specific characteristics of Williams Syndrome, the mechanisms underlying certain behaviors, and even a child's normal development allows us to gain a comprehensive understanding of each situation.

Careful analysis of each individual situation remains essential for us, as is the way each new person is welcomed into a family that already has its own dynamics and characteristics.

We hope this work can be of concrete help to all new parents, not only in finding new solutions to real problems, but above all in discovering their child in all the peculiarities, peculiarities, and characteristics that make him or her special and unique.

# ANALYSIS OF SLEEP QUESTIONNAIRES

The questionnaire aims to begin to organize the many stories we've heard and experienced with families. Thanks to the collaboration with the Italian Williams Syndrome Association (Associazione Italiana Sindrome di Williams Toscana Onlus), it was distributed to 42 families with a member with Williams Syndrome.

It is therefore important to specify that the questionnaire was administered at different ages in the life of the person with Williams Syndrome (child, teenager, or young adult), even though the questions referred to the first 36 months of life.

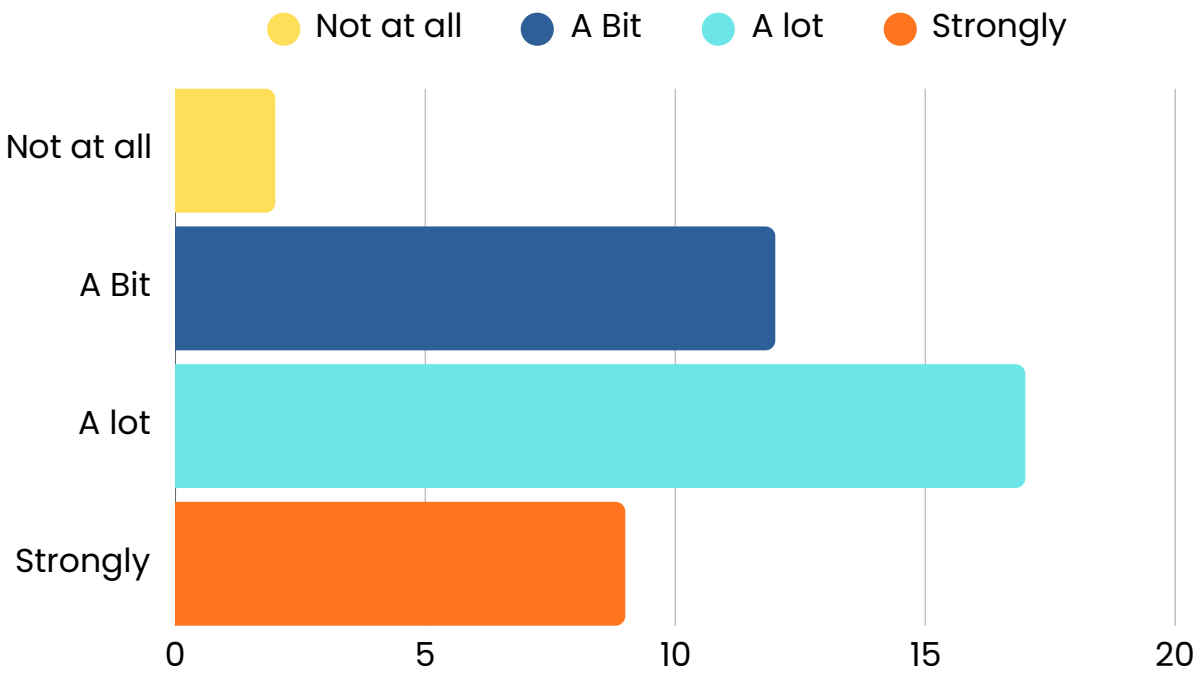
We then asked the "older" ones to recall the early stages of their lives, discovering that it was not difficult to remember that particular period, a clear sign that there were real difficulties, difficult to forget.

Below are the details of each individual question, with the possible answers to choose from, as well as notes on the parents' free stories.

Each question is followed by a simple graph to highlight the percentages. Please note that some questions allowed multiple answers, but not all questionnaires were completed in all fields.

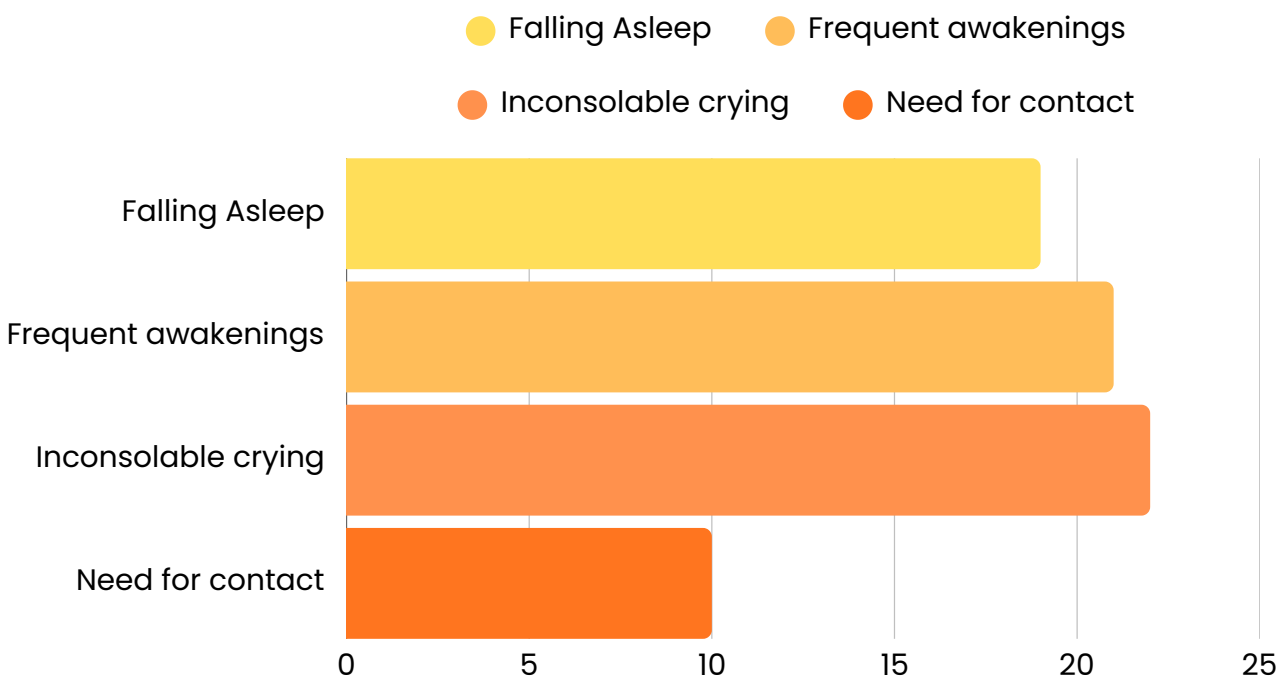
**D1) How much has night-time sleep management affected family balance?**

The first question sought to highlight how the experience of sleep disturbances affected the family's daily routine. Forty out of 42 families responded to this question. Specifically:




We therefore see that for 26 families the lack of continuous sleep had a significant to significant impact: as much as 65%.

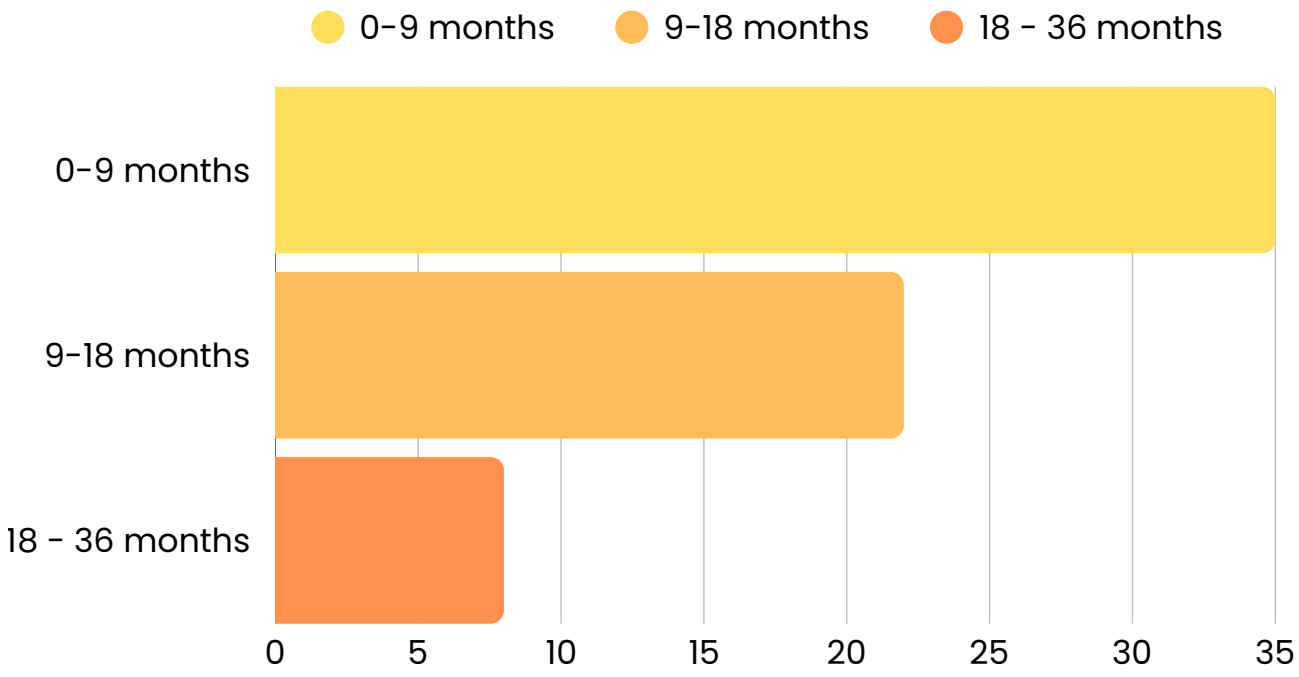
**D2) Which aspects were the most critical?**




30% of the children showed inconsolable crying (22), 29% had frequent awakenings during the night. 26.4% had problems falling asleep.

**3) In which period did you encounter the greatest difficulty in these aspects?**

This question had multiple-choice answers. Some children continued to experience sleep disturbances until age three, uninterruptedly.




However, only 12% of children still have problems in their third year of life.

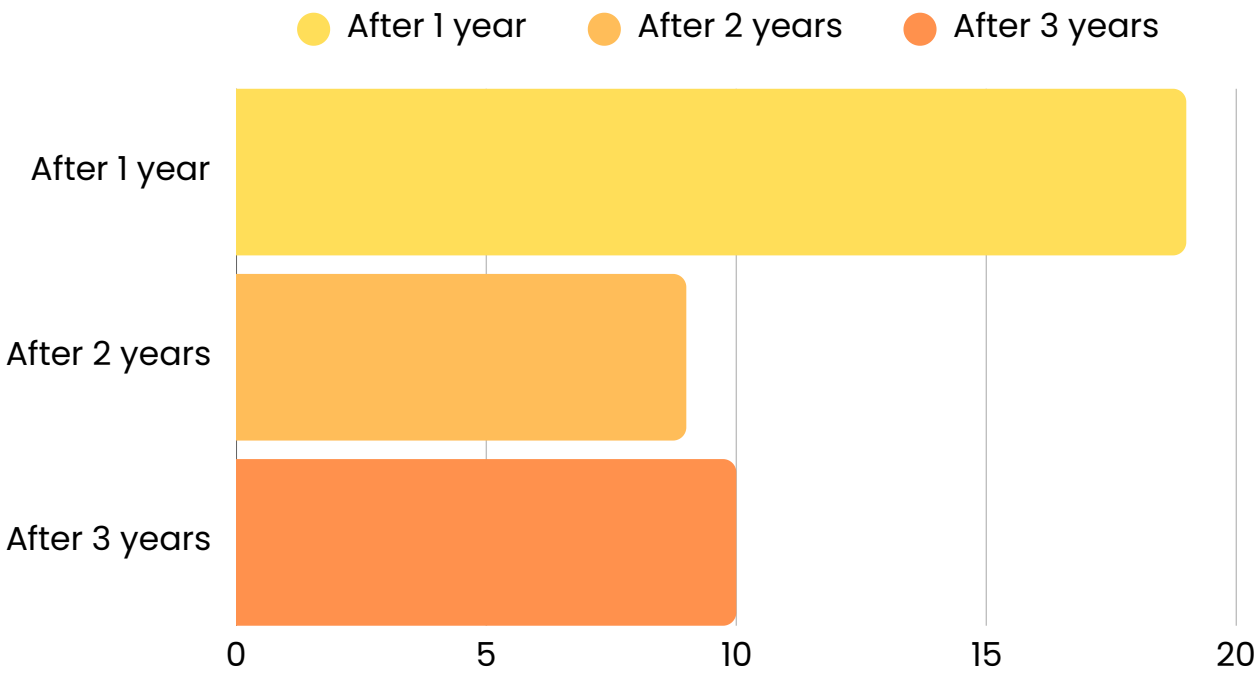
**4) What tools did you adopt to face the difficulties?**

To help their child sleep, parents generally used every means at their disposal, including the use of Noprom under medical supervision. Some parents spent part of their nights in the car, driving around the streets, as a last resort to address the problems they encountered.

We report all the solutions suggested by parents, along with the number of families who have tried them, to show that there are no limits to parental inventiveness.
5 use of Noprom;
3 use of syrups and herbal teas;
1 chamomile with bay leaf;
2 melatonin;
4 make him sleep in the bouncer;
2 in arms;
4 in the stroller;
4 in the car at night;
1 pouch;
5 talk about swings;
3 need for physical contact;
1 uses music;
1 use the hair dryer;
1 used sleep aids;
1 establish a bath-feed-sleep ritual;
many talk about the need for patience, cuddles, and parental shifts;
6 bring the children into the bed (1 due to fear of noises);
1 exchanged day for night.

**5) When did your night sleep begin to become regular?**

As highlighted above, 50% of children begin to normalize their behavior between the ages of one and two. But just as many have problems that persist between the ages of two and three. Approximately 25% persist beyond this age.




However, only 12% of children still have problems in their third year of life.

# SOME OBSERVATIONS ON THE QUESTIONNAIRES

The first years of life of a child with Williams Syndrome are strongly characterized by both significant difficulties in falling asleep and difficulties in maintaining sleep continuously and for a sufficient amount of time to allow for rest.

This affects the quality of life of the child himself, but also that of the entire family, and in particular of the parents who often find themselves having to invent solutions or remedies to alleviate this discomfort.

Starting from the analysis of the results of this questionnaire, we went on to investigate the possible causes, starting from an understanding of the typical characteristics of Williams Syndrome, but also taking into account the normal development of the sleep-wake rhythm in the first years of life.

This work has allowed us to collect parents' responses, which have become a list of suggestions and possible solutions to make this special moment less tiring.

This makes it possible to support parents in addressing these difficulties, while also providing the perspective that all of this is limited to a specific period and can be overcome with the appropriate measures and assistance.

This small study could be the starting point for further investigation and scientific research to arrive at increasingly targeted solutions.

The developmental neuropsychomotor therapists of the Davide e Golia Association

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# WHAT IS WILLIAMS SYNDROME?

Williams syndrome (also known as Williams-Beuren syndrome), whose recognized cause is a microdeletion of chromosome 7, is a multisystem disorder identified as a distinct clinical entity in 1961.

It is therefore a rare, non-degenerative genetic disease, occurring once in 10,000 births. It is present at birth and affects both males and females equally. It is not hereditary and is not caused by medical, environmental, or psychosocial factors, but rather represents chance.
SW affects several areas of development including cognitive, language and psychomotor development.

The peculiar characteristics of Williams Syndrome are linked to:

- Facial Features
- Cardiovascular anomalies
- Neonatal hypercalcemia
- Psychomotor and cognitive development
- Growth
- Genitourinary anomalies
- Ocular anomaly
- Dental anomalies
- ENT anomalies
- Connective tissue and skeletal abnormalities
- Skin abnormalities
- Endocrine-metabolic system

Cognitively, there is great variability among individuals. Some children exhibit average or slightly below-average intelligence levels associated with learning difficulties. Many children border on mild retardation, and some are moderately retarded. Specifically, most children exhibit significant differences in the development of skills across different learning domains.

Children with WS are typically sociable and display strong nonverbal communication from infancy: they use facial expressions, eye contact, and possibly gestures to communicate. They begin speaking later than expected, and despite the extreme variability of cases, they only begin speaking using single words after 18 months. They may demonstrate ease in learning songs, demonstrating good auditory memory and a musical sense. Many children with WS begin to form sentences at around 3 years of age, and language improves and continues to evolve from age 4 or 5.

# APW ITALIA - WILLIAMS SINDROME PEOPLE

How can I get more information about children with WS?
In addition to the content of this brochure, the Associazione Persone Williams Italia Onlus (www.apwitalia.org) is available to provide further information about Williams Syndrome. In addition to the information on the Association's website, a newsletter is published periodically, and updates are also available on the Association's Facebook page.

The Williams Syndrome Association of Italy was founded in 2016 to protect the interests of people with the syndrome and their families.

Its main goals are to support people with Williams Syndrome and their families and to raise awareness and improve knowledge about the Syndrome.

Associazione Persone Williams Italia


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