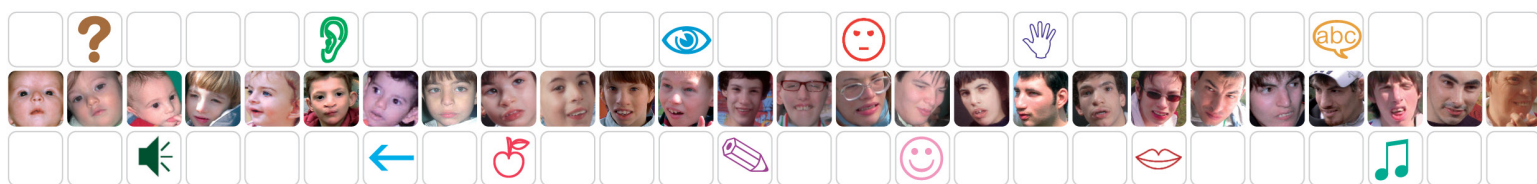


THE CRI DU CHAT SYNDROME TECHNICAL ASPECTS AND EDUCATIONAL GUIDELINES

Edited by Serena Nardi



A.B.C.

"The cri du chat syndrome - technical aspects and educational guidelines".

This is the first edition translated from Italian to English language - 2014

This is a project by A.B.C. Associazione Bambini Cri du chat (Italian Cri du chat Support Group - www.criduchat.it)

Translation edited by Georgiana Ward-Booth from *"La sindrome del cri du chat - Aspetti caratteristici e linee guida educative"* - First edition in Italian language in 2009.

Publication design by Daniele Cavari

© All rights on this publication are reserved; reproduction of text and images is permitted exclusively for non-profit research purposes only, upon previous requests submitted to the A.B.C. Office and with the consent of the author.

This publication has been produced in collaboration with the Fondazione Monte dei Paschi di Siena, which, for several years, has been following and sustaining the Association in its research projects. To them we give all our gratitude.



The English translation of this publication has been realized with the contribution of Autostrade per l'Italia s.p.a. that has supported the Project "ABC Cri du chat Database" for sharing information and guidelines relating to Cri du chat syndrome. To them we give all our gratitude.

autostrade // per l'Italia

*“Come andò che Maestro Ciliegia, falegname,
trovò un pezzo di legno che piangeva e rideva
come un bambino”.*

*“How come that Master Cherry, The Carpenter,
found a piece of wood that wept and laughed
like a child”.*

From ‘*The Adventures of Pinocchio*’

by Carlo Collodi

Dr. Serena Nardi

Graduated in 2001 in Speech Therapy at the School of Medicine and Surgery at the University of Florence. She worked in Florence at the Neurological Clinic of the Careggi Hospital and thereafter at the Meyer Children's Hospital.

She then engaged with A.B.C working closely with children affected by the syndrome, coordinating educational, therapeutic and scholastic projects throughout the country.

In 2006 she obtained the title of Counsellor - Facilitator in the relationship of help.

Serena Nardi participated in numerous training courses further investigating issues related to the developmental age.

At present she provides lectures and training courses and lives and works in Bologna.

This work is dedicated to all those who are dealing with this rare disease, the Cri du Chat Syndrome (SCDC), and need to know a little more about it.

The book is the result of an experience that started some years ago, which had as its primary objective the training of a therapist within the scientific team of the association. A therapist that, for the first time, would have specialised in this rare genetic disease.

Afterwards, the second objective was to concretely start the therapist's work on the field. A long series of home visits to the residence of the families with Cri du Chat children begun with the aims of assisting parents, various professionals involved in the children's education and teachers. As well, the project was able to initiate a data collection on the intellectual, motor, language and especially behavioural development of Cri du Chat children.

The therapist conducting the first and second phase of this challenging project was Dr. Serena Nardi, who was trained with the indispensable help of Dr. Marilena Pedrinazzi, Dr. Luisa Maddalena Medolago Albani, Dr. Luigi Sangalli and under the supervision of Prof. Paola Cerruti Mainardi. The result of this work, which lasted three years, has been summarised in this publication in the form of a "manual" providing important observations on the syndrome and useful information for us parents and for the people working with our children. This data, always associated with scientific methods, are the foundation for future updates and improvements, as the research will move forward.

The contribution made by all members of the A.B.C. Scientific Committee to this project, since its launch, has been unquestionably great and they all deserve our greater gratitude. Those who have directly experienced the presence of the therapist in their homes are well aware of the usefulness of this project and will now be able to better understand some passages through reading this straightforward volume. New parents can instead enjoy the experience encountered by others in the past and start one-foot head.

A heartfelt thanks goes to the Fondazione Monte dei Paschi di Siena that has been supporting us in our projects and that has made this publication possible. Moreover huge gratitude goes to all those local authorities, in which territories some of our Cri du Chat patients lives, who believed and invested in our work also by donating some contributions. Many special thanks to the various administrations of the City of San Casciano in Val di Pesa that since 1995, have always been at the forefront in supporting the projects of the A.B.C. both economically and morally.

Last but not least, my gratitude goes to all those families that, moved by the same desire and need to learn more about their children, have offered so much of their personal experiences to complete this manual.

To them also goes a big encouragement to further pursue working with their children to neutralise, as much as possible, the damages caused by this rare disease.

*Maura Masini,
Mother of Timothy
President of the A.B.C. Associazione Bambini Cri du Chat*



The **A.B.C. Associazione Bambini Cri du Chat** support people with Cri du Chat Syndrome and their families, is the Italian Cri du Chat support group. A.B.C. is a no-profit organisation founded in October 1995 thanks to the commitment of some parents. The association aims to disseminating information to families with children affected by the Cri du Chat syndrome in order to ensure an adequate update on new scientific and therapeutic knowledge and findings, thus ending their feeling of isolation. Furthermore, the association's goal is to raise awareness on the existence of the syndrome and its related problems. Specifically A.B.C. is committed to encourage awareness of physicians, pediatricians, health organizations and educational institutions, therefore improving the care for the affected children. Equally important is the work pursuing the correlation of data linked with the monitoring of the growth and evolution of people affected by SCDC -Syndrome of The Cri du Chat-. In this respect, for several years A.B.C. has been offering a scholarship in genetic-clinical research and has also collaborated on other research projects with Telethon Italia. Additionally, the association has established collaborative relationships on research and therapy development with Orphanet and other institutions and associations.

ABC board of directors 2014

Governing Council:

Maura Masini, *president* - Manuela Barbini Sfondrini, *vice president*.

Members: Dr. Andrea Guala - Giuseppe Galeone - Silvano Borin - Giovanni Boin - Sandra Giani - Luciano Bencini - Massimiliano Pescini - Matteo di Peco, *treasurer*.

Scientific Committee:

Dr. Andrea Guala, *president* - Prof. Paola Cerruti Mainardi, *honorary president* - Dr. Carlo Cappelletti - Dr. Franca Dagna Bricarelli - Dr. Maria Elena Liverani - Dr. Luisa Maddalena Medolago - Dr. Marilena Pedrinazzi - Dr. Mauro Pierluigi - Dr. Luigi Angelo Sangalli - Dr. Fabio Tognon.

Contacts

Head office: Via Machiavelli 56 - 50026 - San Casciano in Val di Pesa (Florence) Italy

E-mail: abc@criduchat.it

Internet site: www.criduchat.it

INDEX

GENETIC ASPECTS	1
SOMATIC AND MEDICAL ASPECTS	3
FEEDING	4
SLEEPING DIFFICULTIES	6
DEVELOPMENT OF THE BASIC PERSONAL AUTONOMIES	7
PSYCHOMOTOR AND LANGUAGE DEVELOPMENT	8
HYPERACTIVITY AND ATTENTION DEFICIT	9
SENSORY ASPECTS	10
ORAL MOTOR FUNCTIONS AND DENTAL / HORTODONTIC PROBLEMS	13
MANUAL SKILLS	14
SCHOLASTIC LEARNING	15

EDUCATION WITHIN THE FAMILY AND THE REHABILITATION PROJECT

BEHAVIOUR MANAGEMENT	16
GENERAL GUIDELINES FOR PARENTS	17
1. Education	17
2. Creating a facilitating environment	20
3. Encouraging autonomy	21
4. Researching for and participating in rehabilitation programs	22
5. Preventing and treating	22
CARE GUIDELINES	23
DIRECTIONS TO THE SCHOOL, PRINCIPLES AND PROCEDURES	24
GUIDELINES FOR THE REHABILITATION PROJECT	26

EVALUATION OF THE CHANGES AND DEVELOPMENTS INDUCED BY THERAPIES IN A SAMPLE OF THIRTY ITALIAN CHILDREN AND TEENAGERS AFFECTED BY THE CRI DU CHAT SYNDROME: MAIN FACTORS

METHOD	28
RESULTS OF THE FIRST EVALUATION	29
THE FIRST EVALUATION: SOME OBSERVATIONS	30
RESULTS OF THE SECOND EVALUATION	33
DATA INTERPRETATION	36
APPENDIX 1 - <i>Some usefull advice</i>	39
APPENDIX 2 - <i>A case report</i>	41
REFERENCES	46



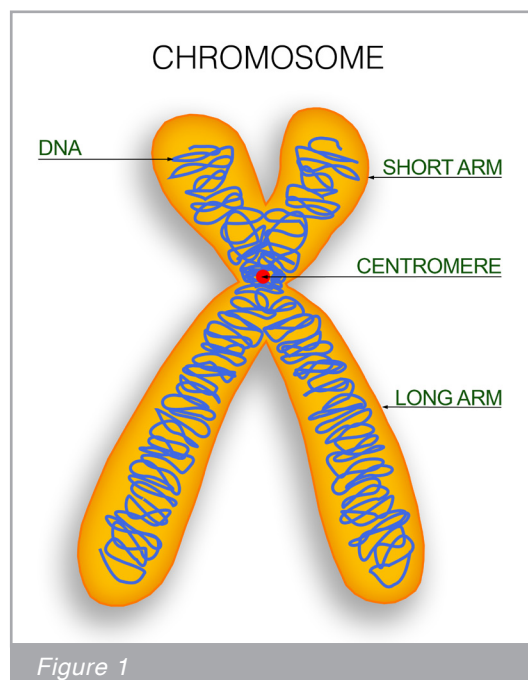
GENETIC ASPECTS

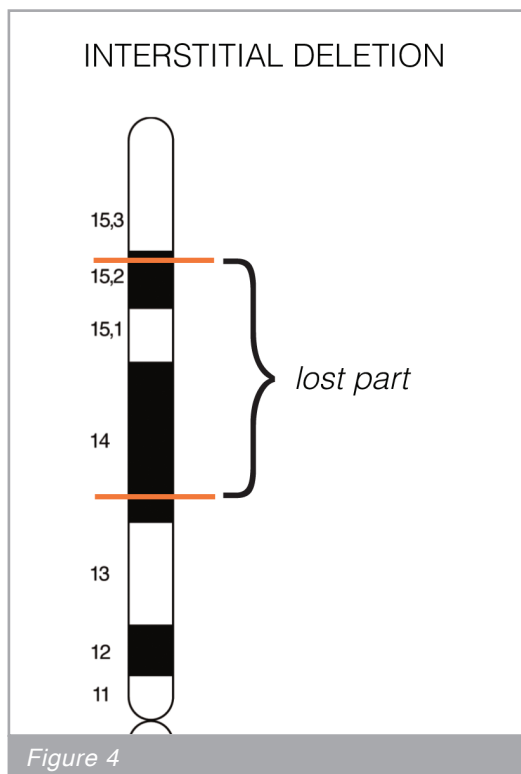
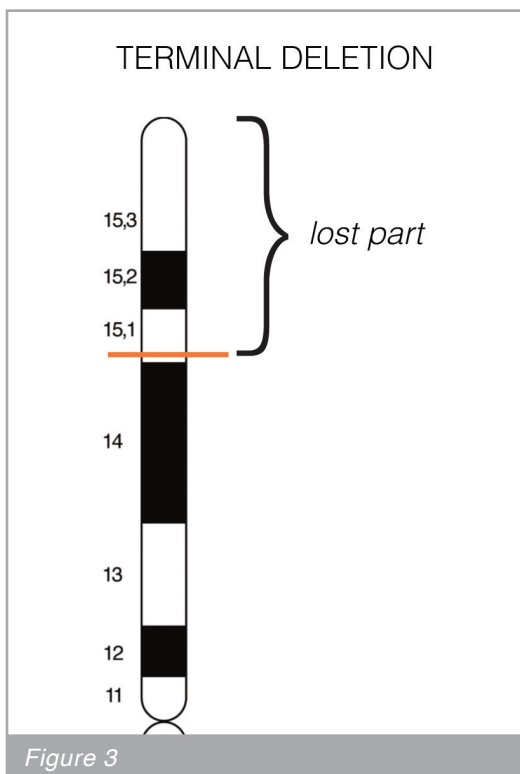
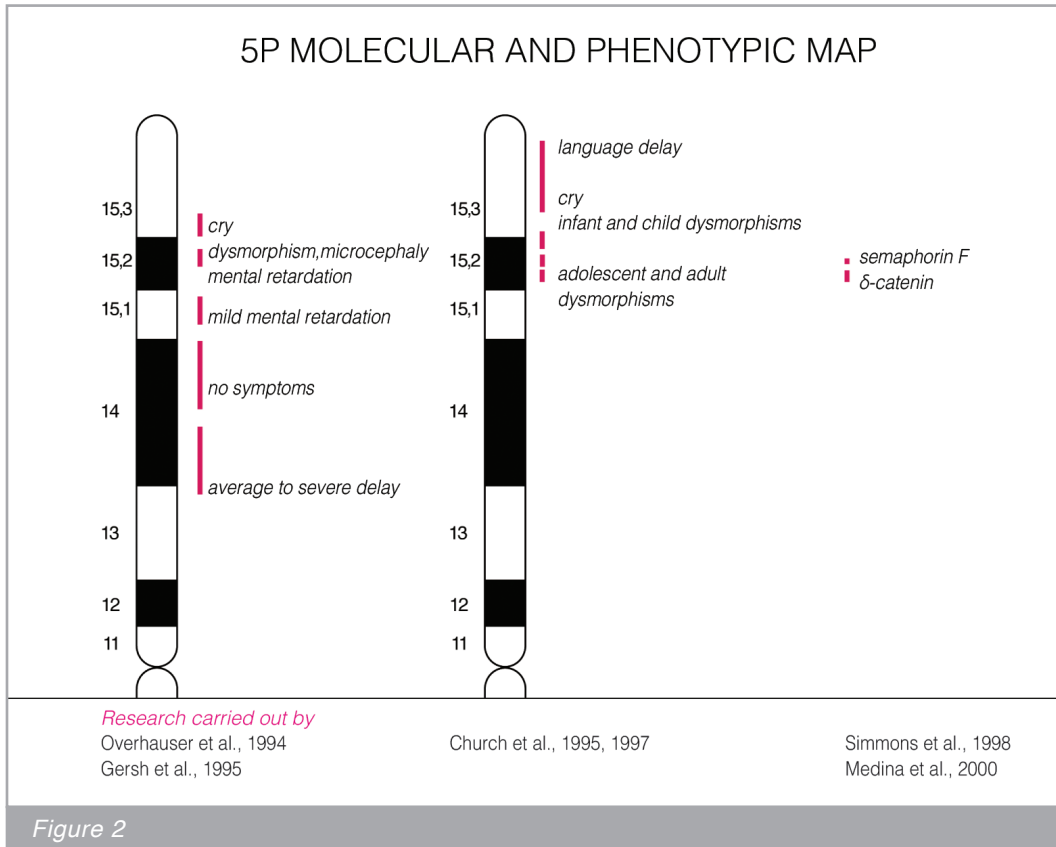
The Cri du Chat syndrome is a rare genetic syndrome discovered and defined in 1963 by the French geneticist Lejeune. The syndrome is caused by a deletion (loss of a fragment) of the short arm of one of the two chromosomes number 5 (5p). (see Fig.1)

In most cases, the deletion is caused by a de novo mutation, thus the parents' karyotype result is normal. In 10-15% of cases the karyotype of a parent may carry a chromosome abnormality, often a balanced translocation, which does not result in any manifestation in the parent but can cause the deletion in the child. In rare cases, the deletion originates from other chromosomal aberrations such as mosaicism, or reverse ring chromosome. Cytogenetic and molecular analysis confirms the presence of critical areas within the short arm of the chromosome 5, the loss of which would be responsible for the pathology typical of the syndrome (see Fig.2). The severity of the syndrome's implications is variable and partially correlated to the extent of the loss of genetic material. So far three genes have been identified two of which, semaphorin and δ -catenin, seem to be involved in the neurological development, and the third, hTERT, which could contribute to phenotypic changes [cf. 27,28].

Implications of the loss of genetic material include: somatic alterations, medical problems, neurological problems, retardation in growth and in psychomotor and language development, with a great differences from child to child.

The deletion can be "terminal" - with a breaking point that determines the loss of a fragment of the chromosome (see Figure 3); "interstitial" - in this scenario the loss happens in a central part of chromosome arm and the break points are two (see Figure 4). Also, in a lesser number of cases, a translocation may occur. A translocation involves the transfer of material between two or more different chromosomes. The balanced translocations are a sort of "equal exchange" of fragments between different chromosomes, which do not involve loss of genetic material and do not cause clinical abnormalities. In unbalanced translocations, one or more chromosomes, have suffered the loss of genetic material, while others have gained a surplus of it. This last type of translocation may occur because the karyotype of one of the parents already contains a balanced translocation. In such situations the clinical condition is usually more severe and the clinical aspects and the somatic features may differ from those characterising the Cri du Chat syndrome. [cf. 3,6,17,18]







The risk that the syndrome manifests itself in a second child is negligible in the case of a de novo mutation, while the possibility increases to 9-19% in the case of a translocation already existing in one karyotype of the parents. Genetic counselling is an essential tool to obtain all necessary information.

However, beyond genetics, a healthy environment of an early stimulation through a targeted therapy, can significantly modify the evolution of the psychomotor development.

SOMATIC AND MEDICAL ASPECTS

CHARACTERISTIC FEATURES AT BIRTH

The following physical characteristics are present in newborn babies with Cri du Chat syndrome (SCDC), and these allow, through clinical observation by medical personnel, an early identification of the syndrome (which must be confirmed by laboratory tests).

The most common traits presented in children with SCDC are [cf. 1]:

- Typical “meowing kitten cry” (due to abnormalities of the larynx: smaller or diamond shaped, limp or hypotonic epiglottis, which all determine a sharper tone of voice)
- Low weight, reduced height, and microcephaly (head circumference below the norm), potential delay of prenatal growth
- Round face with full cheeks
- Root of the nose enlarged
- Low-set ears
- Hypertelorism, epicanthus, down-slanting palpebral fissures, strabismus
- Microretrognathia (small jaw)
- Down-turned mouth
- Dermatoglyphics anomalies, single palmar creases
- Hypotonia

More rarely these traits can be observed:

- Breathing problems (cyanosis, asphyxia) present mainly during the first month of life.
- Feeding problems because of difficulty swallowing and sucking. These problems are usually managed by the departments of neonatology and occasionally by the Neonatal Intensive Care Units (NICU)
- In the early years of life rather frequent respiratory tract infections . For this reason entering nursery may not always be advisable
- malformations (especially renal and cardiac) and less severe malformations such as syndactyly, inguinal hernias, developmental dysplasia of the hip (DDH),
- talipes equinovarus (TEV), hypospadias, cryptorchidism (undescended testes).

In any case, the medical personnel will provide information for therapeutic interventions or specific for screening examinations to be performed.



EVOLUTION OF THE CHARACTERISTIC FEATURES IN LATER YEARS

The physical characteristics change with the passing of years and in older children is commonly observed:

- Narrow and elongated face
- Attenuation of the hypertelorism and epicanthus
- Frequent divergent strabismus
- Prominent supraorbital arches
- Ogival palate (high-arched and narrow palate) and dental malocclusion (open bite)
- Mandibular hypoplasia (small jaw)
- Short metacarpals and metatarsals
- Height and weight generally lower than normal
- Attenuation of the typical cry, maintaining a sharp tone of voice (the larynx remains smaller in size)
- Premature graying
- Hypertonia, which replaces the hypotonia present at birth
- Spastic gait
- More evident microcephaly
- Delay in psychomotor development (acquisition of independent walking, of language etc.).

May also occur:

- Myopia, cataracts and optic nerve problems
- Flat foot and scoliosis
- Very rarely, seizures.

The growth (weight, height and cranial circumference) of a child with SCDC appears reduced compared to the normal standards. An international study has specifically developed growth curves for children affected by the Cri Du Chat syndrome (available online at www.criduchat.it or on the booklet published by A.B.C, *The Cri du Chat Syndrome*, 2002 [cf. 1]). It is recommended to compare the growth of the child with CDCS with these specific growth curves, rather than with the ones referred to the general population (because the two trends are different and therefore difficult to compare).

FEEDING

Feeding difficulties are frequently reported especially in the early years of life. At birth, the sucking may be weak and inefficient; breastfeeding may therefore be difficult. In cases where the child is unable to eat enough, a gastric or nasogastric feeding tube can be used to temporarily support growth. Furthermore, in this first phase of life it may be useful, with the help of a speech-therapist or a physiotherapist, to: determine the most suitable nipples (for both the bottle and the non-nutritive sucking- i.e. the pacifier-), find the best positions to facilitate the feeding and, provide primary orofacial stimulation to facilitate the suction and the swallowing.

Thereafter, coordination difficulties between breathing and swallowing may oc-



cur. These are often manifested in a cough and discomfort when eating and usually happen during the 5th and 6th month of life. Once again, a speech-therapist or a physiotherapist can be a valuable aid in the event that such difficulties are pronounced and frequent. Generally it can be useful to check if the respiratory tract is free and eventually clean the mucus from the nose before starting to feed the baby.

Special attention is required in regard to the of the texture of foods. Generally up to 6 months, the child takes only liquid (milk), and then around the 6th month of life the first homogenised and semi-liquid foods are introduced. Then progressively: smoothies, roughly blended food, and chopped up food are finally introduced to the baby's diet, usually around the 10th-12th month, as well as soft foods cut into small pieces (which dissolve easily in the baby's mouth). With the advice of the paediatrician, these steps can be considered similar in a child affected by CDCS, although each case must be judged

individually. One may also want to introduce the use of a proper glass already after the 12th month of life, so as to gradually decrease the use of the bottle (which should be completely stopped within 18-24 months). Again, a speech therapist can be helpful to assess the competencies of the child and support the parents during the changes of tools, textures of foods and strategies.

This is particularly important in two respects. Firstly, on an oral and motor level, children with SCDC can show some hindrance (see Features gold-motor-orthodontic and dental problems) with difficulties in chewing and managing the "more difficult" food. The resulting protective attitude of the parents is to continue feeding the baby with soft foods for too long, well over their first year of life. Unfortunately, by denying the opportunity to exercise the orofacial muscles for more demanding tasks, this does not facilitate the development of the oral functions of the child. Secondly, on a sensory level, children with SCDC may be more sensitive in their oral, tactile and olfactory-gustatory senses. Therefore it may be the case that the child gets used to certain types of baby food (with some specific taste, odour, colour and texture) rejecting other proposals. Instead, it is important to gradually provide the child with varying stimuli in order to facilitate the development and improvement of his sensory skills. It is thus recommended to make small changes in the consistency, taste or temperature of the food until the child accepts it. We also suggest not to mix many flavours in one meal but divide them into more meals [cf. 15].

Furthermore, allow the child to eat alone with his their own hands very early on, especially when soft but solid foods are introduced to their diet. Additionally, this way offers the child the repeated and motivating opportunity to achieve significant autonomy and exercise his manual and visual skills. These are very important skills that can be extremely difficult to gain through the later work and therapy, which are usually less motivating, thus it is effective. Cutlery can be introduced later on, without any hurry: for a child affected by SCDC controlling a tool is more difficult than controlling his own hand and, the movement that he has to make to lift a spoon or



fork is less demanding, thus less stimulating than the one done by the hands [cfr.1].

The diet and the food education of the child is normally experienced by parents as a very important aspect, especially in the early years of life. Parents of children affected by SCDC may experience an amplified concern because the growth of the child can be slower and because of the difficulties that may arise in the feeding process. Consequently, the support of the paediatrician and speech-therapist to guide the parents in the choice of food and proposals to encourage the development of eating skills of the child is fundamental.

SLEEPING DIFFICULTIES

Parents frequently report difficulties encountered by SCDC children in the sleeping process. We can consider two different issues: the first can be defined as a difficulty in the self-regulation of the sleep-wake cycle. It mainly affects infants and young children and it manifests in difficulties falling asleep and/or with frequent nighttimes and early mornings awakenings. The second, often the result of the first, can be defined as a disorder of the behaviour related to sleep. It is usually reported in older children and teenagers when, in addition to the difficulties of self-regulation of sleep, there are also existing behavioural problems (e.g. They fall asleep and sleep only in the presence of a parent).

Regarding the first aspect, we should remember that often, for newborns, it can be difficult to fall asleep and that they usually learn to do it and regulate it between the third and sixth months of life. The attitude of parents and some small strategies can help the child organise the sleep-wake cycle and facilitate falling asleep [cf. 14].

1. **Rhythms:** it is useful to always put the baby to bed around the same time (between 20.30 and 21) and wake them up in the morning at a set hour (and not let him-her sleep for a long time in the morning because he-she barely slept at night). As the rhythm will regularise, the child him-herself will wake up the morning after having slept enough.

2. **Routines:** the rituals of sleep are very important: a lullaby, a goodnight kiss, tucking the covers etc.. All these repeated small acts contribute in creating an atmosphere of security that reassures the child and makes him-her feels protected.

3. **Let the baby cry:** we advise parents not to rush to reassure the child every time they cry but to give them time to learn how to self-consoling. This can be heart-breaking for some parents, in which case we recommend a gradual decrease (but constant) of their presence. In the first case the child will learn to sleep in a few days, while in the second case the time will be a bit longer.

4. **Constance:** it is very important to maintain the same methods constantly (even if your child has a cold!).

5. **Attitude and mood of the parents:** remember that infants and children are very sensitive to emotional states and tension. if the bedtime routine is accompanied by



anxiety and tension by the parent, the child may have more difficulty to relax and fall asleep.

6. **Activities during the day:** particularly useful are outdoor activities.

Unless there are special medical problems reported, it is recommended to let the baby sleep in their crib from the beginning (possibly the use of intercoms, baby-cry monitors, or similar equipment can reassure the parents that everything is going well).

The second aspect is visible in most of the grown ups who did not receive a sleeping education. In these cases the main problem is that they are dependent on the presence of a parent, often the mother. The child falls asleep only in the bed of the parents forcing one of them to sleep somewhere else. It's not too late to teach your child to sleep alone but it will require more time and effort.

DEVELOPMENT OF THE BASIC PERSONAL AUTONOMIES

Basic personal autonomies include the ability to eat alone, sleep alone, undressing and dressing, sphincter control (at day and night), knowing how to go alone to the toilet.

Many SCDC children easily reach these basic autonomies (possibly requiring little aids from the adults) if they are educated from an early age. It is more likely that these basic autonomies are absent in Cri du Chat grown ups who have not been educated in this respect, often due to a lack of specific instructions and information offered to the parents from doctors and rehabilitators.

Parents are urged to pay attention to these issues from the early days. In particular, as previously said an education aimed at achieving independent sleeping habits in care institutions should begin almost immediately (see par. 4). Moreover, since the introduction of first solid foods in the children's diet, you can help and facilitate them to feed themselves autonomously with their hands (see par. 3). According to child development, generally between 2 and 3 years, you can begin to teach them how to dress and undress, introducing gradually more demanding procedures. To begin with it may be just asking the child to pull down the t-shirt or jumper after it has been put over the head and arms by the parent, or to slip one arm after the other in the t-shirt etc. till the development of complete autonomy. It should also be kept in mind that undressing is easier than dressing and that the child may need some help to complete activities involving fine motor skills. For example, tying and untying the strings of tennis shoes may be too difficult and require the intervention of an adult, while closing or opening the Velcro strips can be done by the child on their own.

In regard to the sphincter control, it may be useful the advice of an experienced rehabilitator or a child psychiatrist. In particular it may be proposed to make the child use the toilet when they are able to hold the sitting position with sufficient balance but possibly before they learn to stand and walk alone. It can be useful to make your baby sit for one or two minutes on the toilet after meals to facilitate for the child



the association between the bathroom and the defecation (a full stomach determines the reflection of intestinal emptying). Then, you should always put the child on the toilet (with possible adjustments to make the sitting comfortable) at regular intervals of about 30 minutes, leaving them without nappies all day long (this work is better suited to be done in spring-summer). The familiarisation with the toilet and the possibility to remove the nappies is usually gained with this type of work even though the timing may be different from child to child.

PSYCHOMOTOR AND LANGUAGE DEVELOPMENT

The psychomotor development is severely delayed in Cri du Chat children, although recent researches highlight more positive and optimistic results than in the past [cf. 19, 20]. In particular, fundamental (or crucial) are the modes in the education and upbringing of the child. Early research refers to children living and growing in care institutions albeit with a delay while subsequent one was based on the observation of children raised by their own family and which were early introduced to therapeutic projects. The latter have shown a better cognitive and psychomotor development [cf. 6, 8]. This already suggests that the severity of the genetic damage is not the only important factor and that instead the environment that they grow up in plays a crucial role in the development of the child.

In the 2001 study of Mainardi and collaboration [cf. 6], after having introduced 80 Cri du Chat children to the scale of Denver, it was found that 50% of the children were able to stand up and support themselves alone within the 21st month of life and were able to walk alone at 3 years (and 95% walk within eight years). Furthermore, 50% of children, within the 9th month were able to grab objects and at four years old able to eat alone. 50% also said “Mum and Dad” at three and could combine two words at five years.

With a few exceptions, Cri du Chat children will walk independently albeit with a delay in the acquisition of this competence. Therefore, the priority of the physiotherapy intervention should not be the verticalisation (e.g. the use of statics) and the achievement of walking but the promotion of a good motor organisation. This can be achieved through practicing directed movements: sliding, crawling and posture steps are preparatory and fundamental to the development of the ability of walking autonomously, and in a more harmonious way. In fact, the main difficulties are observed in the dynamic coordination of hand movements and fine motor skills.

Regarding the language development, which is particularly compromised in children affected by SCDC, two lines of divergence from the regular development figure are observable [cf. 12]. The first one is the difference between the child's chronological age and the presumed linguistic one. This difference is amplified as the age increases (e.g. The child's chronological age is 6 years but its language can be similar to that of a 2 year old). The other line of divergence is the one between language comprehension and expressive language: in fact, while



the understanding of language appears to evolve till about the linguistic age of 10 years, in most children, it has been observed that expressive language does not proceed beyond the age of 2.3 years. A study of Söhner and Mitchell, 1991 [cf. 24], suggests a correlation between the cognitive and motor delay and that of the expressive language.

Another observation, related to the language and communication skills, which needs to be considered. In some cases the expressive language skills are very limited but is noted a good use of non-verbal communication skills such as the use of facial expressions, gestures and sequences of gestures. This suggests the possibility of using other communication channels (with reference to the principles of the AAC -Augmentative Alternative Communication- or the CF -Facilitated Communication-) to partly mitigate the limitations of verbal communication. In other cases it is noticed little or no use at all of these non-verbal communication modes, alongside the absence of verbal communication. Observing these children, who do not necessarily have the most extensive gene loss, it is highlighted the presence of major sensory difficulties and “poor” and stereotyped behaviour. These, firstly affect the linguistic and communicative access (above all the awareness of the fact that the external environment can communicate) and secondly the possibility of expression (understanding the fact of being able to communicate with the external environment).

These aspects will be further elaborated in the 3rd part of the book.

HYPERACTIVITY AND ATTENTION DEFICIT

Hyperactivity is one of the most significant and more frequently presented consequence of the syndrome. This can manifest itself in various modes and intensities, and it is especially noticeable when the child is more tired or stressed, or seeking the attention of an adult. The hyperactivity is expressed through restlessness, irritability, impulsiveness and inability to wait. Moreover, the child can quickly swop one object or toy with another one, without even playing with it or using it in its proper functions (e.g. not using the pencil to scribble but putting it in their mouth or slams it on the floor). Also, the child can perceive space as if it has no limits and no rules (e.g. the child might run down the street without the perception of danger or move around the house throwing any ornaments on the floor, thus forcing parents to seal doors and drawers or transfer all the furnishings to a highest, inaccessible level).

At the same time it is observable that a low attention and concentration of the child who can be extremely easily distracted by other audio and visual stimuli. Therefore it is very important to create a facilitating environment in terms of avoiding sensory distractions, so that the child can engage in a more continuous activity [cf. 21].

In order to identify some benchmarks in regard to the expected attention span, it is important to consider the alleged mental age of the child. In fact, if the child's



mental age is 2 years, they will be able to keep the focus on one activity for no more than 3 or 4 minutes. It is therefore important that the proposed activity, both within the family environment and the therapeutic one, as well as the scholastic one, are of short duration and are repeated several times during the day, in order to be learned by the child [cf. 11].

For those children showing these difficulties, it becomes very important to maintain regular rhythms in the articulation of daily activities and an adequate nights sleep.

It is also fundamental the role of the adult in maintaining a firm and resolute educational attitude in order to teach the child rules that can guide them in their daily-life routine. It is also important, that the adult establishes eye contact with the child and talk to them in simple sentences accompanied by a mimic congruent with the verbal message (if the aim is to blame them then you shouldn't smile but have an angry face). Besides, the role of the adult cannot do without involving the child in motor and sensory experiences, or activities and exercises that teach and show them a way to direct their energy in a targeted and concretely conceptualised way [cf. 22].

SENSORY ASPECTS

RELATIONSHIP BETWEEN SENSORY ASPECTS AND BEHAVIOURAL CHARACTERISTICS

Hypersensitivity to sounds and images is one of the main characteristics of the Cri du Chat Syndrome [cf. 12, 13]. These hypersensitivities originate from a deficiency of the central nervous system (CNS) at the level of the integration and the processing of sensory information, thus the sensory organs and the decoding device are usually undamaged.

The hypersensitivity to sounds, on the one hand, involves the perception of noise with very low frequency and all those sounds constituting the background noise of the environment (which are usually perceived only if they become conscious object of our attention). On the other hand, great intensity noises are amplified and they become of large annoyance for the child, especially if they are unpredictable. The consequence is that the child will be particularly attracted by barely perceptible noises, usually with no significance, and will demonstrate more difficulty in selecting, among all the simultaneous noises, such as the human voice.

To deal with the language deficits is, therefore, necessary an evaluation of these aspects and their possible treatments. The hypersensitivity to sounds may occur with different modes and intensity. Children may raise their attention to a simple creak, or be restless in environments with loud background noise, as well as being distressed and frightened by loud and sudden noises (e.g. an ambulance siren, a thunder, a motorcycle, etc...). It is also very common to see the child, with a hypersensitivity to sounds, turning up the music volume. This fact often leaves the parents dumbfounded. Yet, it must be considered that the music has a rhythm,



not a sudden burst, thus it contains a very valuable feature for the child: it is predictable. Consequently, loud music can be greatly enjoyed by the child, not only because it may be pleasant, but because it can cover all other ambient sounds that are much less predictable.

Characteristics of the syndrome, although different from case to case, are the alterations of other sensory channels. The next section lists the major behavioural manifestations attributed to these sensory deficits. As for the tactile sensitivity, certain alterations can be noticed: the sensitivity to pain is usually reduced so that the child does not cry, or cries just a little bit, in case, for example, of a fall. A similar feeling for us may be, after an anaesthesia at the dentist, the way you sense your mouth, your tongue, etc.). In some other cases, even though more rarely, the sensitivity to pain is more pronounced, or it can be perceived after some time. The superficial tactile sensitivity is usually more accentuated. For example, all those superficial and slight contacts, like a caress or a hair comb, can result in being very annoying for the child (to understand this sensation we can think of how much more sensitive is the skin after a sunburn). Paradoxically then, a slap on the arm can be felt as more pleasant than a caress. In other cases, or in addition to these alterations, the child may show such behaviours as scratching, biting and rubbing their skin, often to the point of tearing the skin, as if they had a sort of perpetual annoyance (think for example of the sensation on the skin produced by an insect bite).

Alterations of taste and smell sensitivity have already been mentioned at page 4 under the “feeding” paragraph.

SENSORY DEFICITS AND STEREOTYPED BEHAVIOURS

Sensory alterations can be divided into:

- **HYPERSENSITIVITY** (over stimulation): the sensory pathway is excessively open and therefore, the stimulation that reaches the brain is excessive and troublesome.
- **HYPO-SENSITIVITY** (under stimulation): the sensory pathway is not sufficiently open and as a consequence, the stimulation is too short and it cannot reach the brain
- **WHITE NOISE**: the sensory pathway, due to a defect, produces independent sensory stimuli that do not correspond to what is happening in the outside world. Hence, the message coming from the outside is altered or, in extreme cases, is covered by the background noise of the system. For this type of alterations the cause is not at the peripheral level (e.g. the sense organs do not usually have alterations), but is to be found in an impaired functioning of the CNS.

The stereotyped behaviours that the child may have are related to these altered sensory modalities. [cfr.22]

The root of many stereotypes behaviours and typical attitudes, are sensory alterations. ‘Hyper - alterations’ are characteristic of the SCDC. The sensory channel is too open and the CNS receives a vast amount of information. At the same time, the



CLASSIFICATION OF STEREOTYPED BEHAVIOURS AND TYPICAL ATTITUDES BASED ON THE DIFFERENT ALTERED SENSORY MODALITIES

(Extract from: Luigi Sangalli, '*L'attività motoria compensativa*', Trentouno Edizioni, 2006)

Alterations of the tactile sensory perception correspond to the following stereotyped behaviours:

- Walking on tiptoes
- Reduced sensitivity to pain
- Annoyed reactions to caresses or kisses
- Extreme sensitivity to pain
- Presence of a permanent stimulus (e.g. the child continuously rubs themselves where they have hit half an hour early)
- Shaking of the hands and / or slapping on the forehead, face or side of the face.
- Knocking their head against hard surfaces
- Creating deep wounds in their skin
- Slapping, hitting, biting themselves with the aim of injuring themselves

Visual sensory problems correspond to the following stereotyped behaviour:

- Avoiding to establish eye contact
- Staring at their hands, objects or other details in the surrounding environment for at least five seconds
- Quickly moving their fingers or hands in front of the eyes for several seconds or more
- Staring at things in the air that they can only see
- Moving random objects with the aim of making them spin (e.g. plates, cups, glasses etc...)
- Rotating and spinning balls or wheels
- Stacking objects with extreme precision
- Creating wires with their saliva

Auditory sensory problems correspond to the following stereotyped behaviour:

- Emitting high-pitched and / or loud sounds or self-stimulation vocalisation
- Hitting their head or ears
- Banging objects to produce noise
- Isolating themselves in music and / or listening to very loud music
- Covering their ears with their hands

Vestibular sensory problems correspond to the following stereotyped behaviour:

- Rolling themselves over or turning in circles
- Swinging back and forth while sitting or standing
- Moving from place to place with fast and / or with pawing movements
- Jumping as they move from one place to another
- Assuming strange postures, often upside down, while sitting or lying

Sensory problems of smell and taste correspond to the following stereotyped behaviour:

- Eating just some specific foods and refusing to eat others that usually everyone eats
- Eating anything within reach (faeces, soil, detergents, etc...)
- Licking inedible objects (e.g. a person's hands, clothing, floors, etc...)
- Smelling objects (e.g. toys, people's hands, hair, etc...)
- Eating with their face curved toward the plate



sensory / attentional system is not fully operative. The consequence is that the CNS is flooded with information that it can not manage to organise. This produces, in the child, a difficulty of constructing models of representation of themselves and of the external reality.

By observing what the child spontaneously does it is possible to highlight some of these behaviours, thus infer the sensory abnormalities presented.

The evaluation and treatment of sensory alterations is of fundamental importance and it must necessarily be included in the child's educational and rehabilitation project.

Often, but always with great differences from case to case, parents and educators can encounter problematic behaviours that are difficult to manage. Some of these are commonly referred to as self-harm or self-aggressive (aggressive behaviours that the child direct towards themselves), others are instead called hetero-aggressive (when facing other). It is important to keep in mind the connection between sensory deficits and auto / hetero aggressive behaviours [cfr.9].

For example, a girl suffering from SCDC had learned to slam her head every time she was contradicted or something was denied to her. The strategy appeared to be very effective since everyone bended to her will, fearing that she would have hurt herself. But the important point here is that she was not hurting herself! This strategy had for her no cost, because the reduced perception of pain, typical of SCDC, prevented her from feeling the pain. Therefore, this cannot be regarded as a self-harm behaviour, but quite the opposite. In this case, the adults have to ensure that she does not get in a situation of real danger, but at the same time do not please her. Not giving up in front of her blackmail and ignore her behaviour as far as possible. Other times the child may be pinching and scratching themselves repeatedly in the same place, self-causing lacerations and wounds of the skin. Other times the child can hit their ears or be very noisy in their games. These are all sensory self-stimulations and adults need to ask themselves what is missing or what is too much for this child, in terms of sensory perceptions, in order to understand what can be concretely done.

The presence of stereotyped or problematic behaviours is the index of sensory abnormalities, thus it should be treated as such.

ORAL MOTOR FUNCTIONS AND DENTAL / HORTODONTIC PROBLEMS

These are the main oral motor alterations that may be found in SCDC children:

- The tongue is less able to move
- The activities of the various facial and oral muscles are not well articulated
- The root of the tongue is short and stubby
- The tongue appears thin and narrow (as a "cigarette") and sometimes more rigid
- The mimic musculature of the face (which allows small movements
- and facial expressions) is hypotonic
- The muscles of the lips are disharmonic: longer the upper lip,



- shorter the lower one
- Neurological difficulty in coordinating the muscles of the tongue with those of the face (genital deficit)
- Drooling due to altered sensitivity and / or faults in the oral motor control

Structurally it is observed:

- Narrow and deep (ogival) palate
- Teething problems and dental malocclusion (open bite, deep bite, etc...)
- Small jaw (the lower third of the face, as a whole, appears less developed than the upper part. Usually this is referred to as microretrognathia)

Taking into account the tendency to develop these disorders, it is useful to consider several aspects that can facilitate the organisation of the oral functions and reduce dental problems [cfr.25]:

1. Special care in the feeding process: see p. 4 (texture of food / use of bottles and pacifiers)

2. Food: avoiding too many sugary foods or sugary drinks

3. Oral hygiene: it's important to keep clean the mouth and the teeth of the child. Initially there is no need to use toothpaste (which produces a lot of foam that is difficult to wash away) or toothbrush. It is advisable to just moisten a piece of gauze, roll it up on the little finger and pass it over the gums, tongue and palate, and when they appear, on the teeth, moving from the gum towards the tooth. The toothbrush can be introduced later on, around the second year

4. General motor skills: gross motor development precedes and facilitates the organisation fine motor skills (on both an oral and manual level). In particular, activities on their stomach and sliding exercises facilitate the development and organisation of the chewing and then the swallowing

5. Oral motor skills: specific oral stimulations such as facial massages can be introduced relatively early following the instructions of a speech therapist (anyway, in the early days are preferable body massages that exclude the face)

6. Regular dental visits to be arranged with the dentist, from the appearance of the first teeth.

MANUAL SKILLS

Motor skills can be divided into two main categories: gross motor skills and fine ones. It is also understandable how the first will precede the second. All Cri du Chat children usually develop gross motor skills, although times may be longer. What remains difficult is controlling fine movements, which require a very high level of sensory and motor coordination (e.g. movements to articulate the pronunciation or hand writing movements). As with any other skills, to identify and promote the manual skills necessary to identify their prerequisites. For example, a simple and motivating exercise can be to repeatedly make the child eat with their hands.



SCHOLASTIC LEARNINGS

Is generally not recommended to enter the child into a childcare, both for the occurrence of frequent respiratory infections and for the overall immaturity of the child. However, it is usually very positive to enter the child in nursery school, even if they may need longer time to adapt to the different environment. The more positive experiences arise when there is an interaction between parents, teachers and the rehabilitation team. Often the support of child psychiatrists and therapists are useful to define the educational goals and to identify the methods to be used.

The second part of the book will provide more details in regard to the promotion and methods of scholastic learning.



EDUCATION WITHIN THE FAMILY AND THE REHABILITATION PROJECT

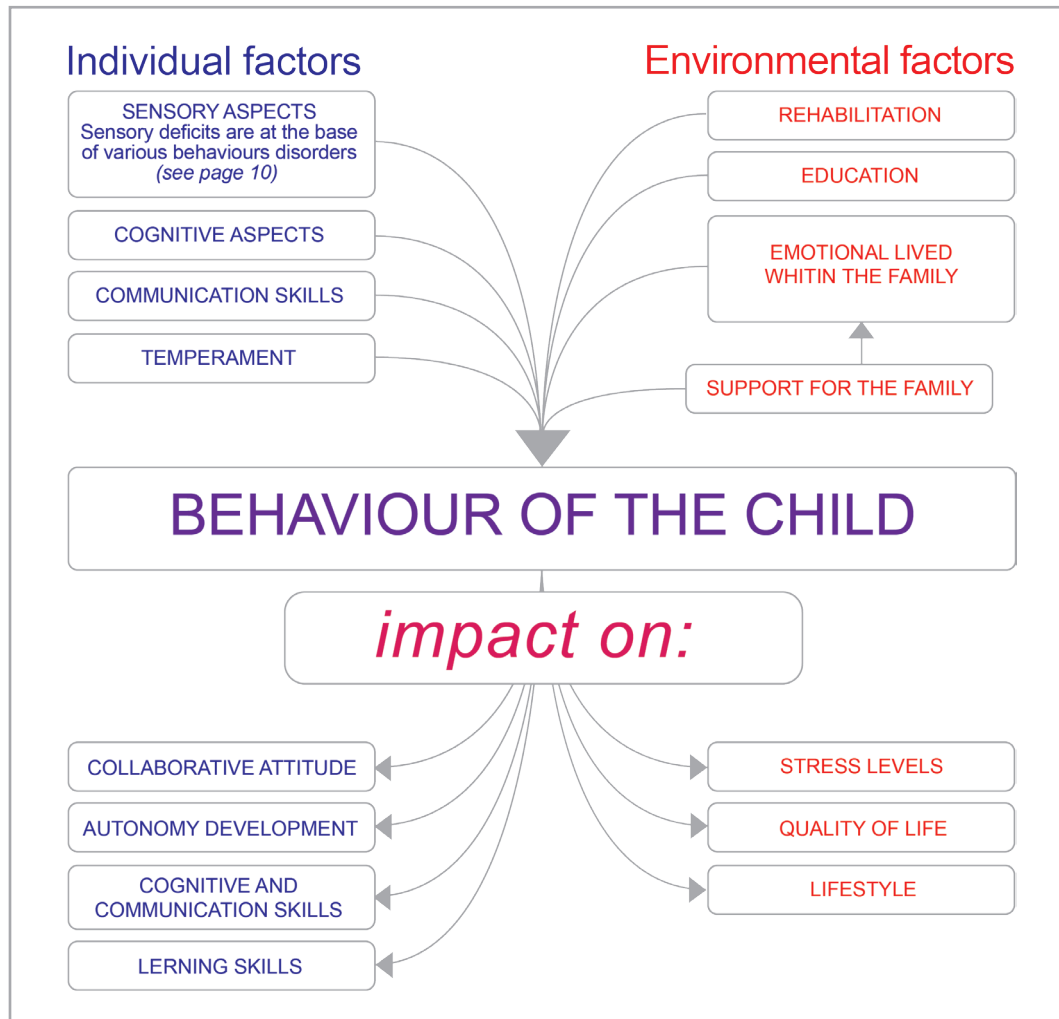
BEHAVIOUR MANAGEMENT

Whilst observing a number of children affected by SCDC it has been noted that one of the major problems for the family is the management of the child's behaviour. In fact, even if these children are usually sociable and affectionate, often their behaviour can be an issue for the family who may wonder which approach is more appropriate in specific circumstances.

Thus, behaviour is a crucial aspect in the evolution of a Cri du Chat child. Two key considerations are necessary:

- 1) The child's behaviour is the result of a number of factors
- 2) Behaviour has an impact on the child's overall development and on the quality of family life

Hence, it follows that the presence of problematic behaviour, the lack of co-operation and the reduced development of basic autonomy in the child will affect and limit their learning ability. This may be because all the attention is focused on



the behavioural disorder and this paradoxically may worsen the condition. Another reason may be that the child is not actually able to engage in the performance of any activity, and-or that they are less perceptive to learning opportunities naturally offered by everyday situations.

Thus, a cognitive limitation originating in the relationship between the individual and the environment is added to the original cognitive limitation caused by the genetic alteration.

GENERAL GUIDELINES FOR PARENTS

The following are brief guidelines. For a better understanding it is helpful to read the first part of the book.

1. Education

Educating a child in itself is definitely not an easy task in life and this is surely



demonstrated by the broad range of books and manuals available on this subject. In fact, this task may presents us with a series of small or significant organisational and practical problems. Educating a child implies making choices for another person, it forces us to constantly question ourselves and our beliefs. Moreover, it can challenge our personal way of interpreting the world, requiring us to change perspective and to look and listen before acting.

Educating a child with a disability requires more effort and involves a more articulated emotional experience. It is important to reflect on this difficulty, to accept it and deal constructively with the reality. This may imply asking for help or looking for “alliances”. For example, talking with other parents of children with special needs can help you to feel less isolated and may provide useful information and ideas. Seeking counselling could also help to focus the difficulties mobilising our internal resources. Carving out personal spaces and free time can help to regenerate and regain energy. Furthermore, actively participating in educational and rehabilitative programs can enhance a sense of confidence and positivity.

In the education of Cri du Chat children, experience suggests us some useful principles:

RULES

Respect of rules is a problematic aspect that deserves special attention. A child who habitually fails to comply with rules, either because the meaning of the rule is not understood or because they intentionally break it to provoke the adult, is often a child bewildered and confused. For a Cri du Chat child this non-compliance results in the child themselves and the family being in a chaotic state which is difficult to resolve.

Rules - Little by little

There is no need to fill the life of the child with negative reproaches and rules. Instead, it is often preferable to set a few, but well-defined, shared and non-retractable rules. Ultimately, when a rule is internalised by the child, there will be no need to reiterate it from the outside because this will already be integrated in the behaviour of the child.

Rules - Shared

An iconic situation is when the mother says, “No stop!” and the grandmother follows with “But poor him, let him do what he wants!”. This situation can create a moment of impasse so that the mother does not know any longer what to do and the child’s behaviour that follows is usually to ignore the rule set by the mother. This is why it is important not to disregard what the mother suggests. If the grandmother did not agree with the rule, it is still important to talk about it and discuss it, but at another time. This problem is also very common between the two parents, between parents and grandparents and often between family and outsiders. The mother can not ask everyone to share her beliefs and decisions, but certainly in the contingent moment she can maintain and defend her position, and this will result as sufficient to enforce the rule.



Rules - Constants

A rule to be learned must be retained, it is important not to give up when tired or exhausted by the child's blackmails.

Rules - Clear

Many words or explanations are not required. Saying "No!", "Stop!", "Enough!" Etc. ... can meet the purpose more immediately rather than saying: "Do not do this because you might get hurt.". Very simple explanations understandable to the child may be useful, but at a later time.

RITUALS AND ROUTINES

What is meant by ritual is a sequence of activities and actions that are repeated over time to accompany the child and reassure them. E.g. the bedtime ritual: to put the baby to bed, turn down the light, sit next to them, singing a song or tell them a short story, give them the goodnight kiss and then leave the room. Even small actions at home can be thought of as giving the child a certain sequential routine so that these actions can be better absorbed and spontaneously reproduced by the child. Another example: the child comes home and with the help and guidance of an adult, removes their backpack, puts it in the right place, removes their jacket and hangs it up, takes off their shoes and finally goes to the bathroom to wash their hands.

RHYTHMS

It can be particularly useful to try and structure the child's day and keep a regular pace. In fact, a regular rhythm helps the child to be more time orientated. This requires them to know where they will be at a specific time and what may happen next, thus reducing their level of anxiety. For example, a typical day may be organised as it follows: breakfast - school - lunch - psychomotor exercises - school - gym - out with the grandparents - snack with the grandparents - home with mum and dad etc... . If this is their typical day, is very important to explain to the child with a few simple words what changes may occur, in order to mitigate the child's anxiety in front of an unexpected event.

EFFECTIVE COMMUNICATION

Congruency

Communication consists of words, facial expressions, gestures, tone of voice... and much of the meaning of a communication is entrusted to non-verbal aspects. If a mother reproaches her child but feels guilty in doing so, she will try at the same time to reassure the child. So, while her voice will say "No!", the indulgent gaze and a smile that emerges or a gentle tone of voice will tell the child "Go ahead!". The child will then continue to do what they were not meant to be doing and they will have learned that the word NO means YES. Therefore, it is fundamental to maintain a congruency between words and non-verbal aspects.



ACTING WITH PERSPECTIVE

It is important to keep in mind two things: firstly that the baby in front of you will eventually grow and secondly that for them it is much more difficult to forget things that they have previously learned rather than it is easy for them to learn new rules. Bearing this in mind, what is a socially acceptable behaviour at the age of three may not be so at the age of fifteen. For example, if the child at the age of three has been taught to greet everyone, hugging and giving kisses, this will be considered acceptable, but if a boy of fifteen years old continues in doing so, this may no longer be an appropriate behaviour. Thus, it is better to teach the child to hug and kiss just family members and close friends and wave and smile to the less familiar people.

Let us remember also that the disabled child or teenager lives in a social context that does not always adapt to their needs, so general rules of politeness that are applied to all children must also be valid for them.

2. Creating a facilitating environment

The first part of this book outlines the various areas of difficulty that a child with SCDC may experience, hereafter we refer particularly to sensory difficulties. From a sensory point of view the environment in which the child lives can be organised and structured to be less annoying and therefore more relaxing and facilitative.

FROM AN AUDITORY POINT OF VIEW

It is important to make sure that there is not too much background noise (e.g. hums, clicks etc...) at least in the room where the child spends the majority of their time. If your child has trouble sleeping, try for example to see if there are any noises at night that disturb them (e.g. the water flowing through the radiator). Some children may sleep just whilst they can hear familiar ambient noises, but may wake up when these noises are missing and other strange noises arise, which can be a source of fear.

Please remember that the more outside noises there are, the more noise the child will make (to re-direct their attention to the sound produced and isolate themselves from the other noises). Therefore, it is always useful to distinguish and eliminate the auditory stimuli that may distract the child's attention, e.g. switching the TV off whilst talking to the child. Even the induction to school can be difficult precisely because the school environment is a very loud and disturbing one. It is therefore important to implement a series of small changes even within the school environment (see part 2 - Directions to the school). In general, to attract the child's attention, it is useful to speak with a low and soft voice almost whispering. It is also advisable to speak with a calm voice with no overhangs (i.e. avoid raising or lowering the intensity or tone), avoid yelling (because this impedes the understanding of the language: the child may be attracted by the acoustic stimulus, but will not listen to the words). Thus, it is recommended to avoid places which are too chaotic and instead offer the child a small and quiet environment. Additionally, it can be useful talking to the child with simple



messages, using few words and trying to establish eye contact with them. Last but not least, if there are any noises that may frighten the child (e.g. the vacuum cleaner) it is useful to warn them in advance.

FROM A VISUAL POINT OF VIEW

Cri du Chat children should not be simultaneously exposed to too many visual stimuli. Often babies' rooms are full of toys colours, patterns, etc... . In such an environment, SCDC children will switch their attention from one thing to another in a chaotic way. Visually, the environment should be relaxing and basic, for instance with all the toys placed inside a box and with reduced colours, objects and drawings around the room. It can be helpful to present the child with one toy at a time and put it away as soon as they finish playing with it. It can also be helpful that the room in which the child spends majority of their time is not too bright but out of direct sunlight. Even when concerning the choice of toys it is recommended to keep these suggestions in mind: for example a toy that lights up, talks and moves may be too much from a sensory point of view and too little from a cognitive point of view. Therefore, an illustration book should not be too full of drawings, because the child will focus on a particular part of the design and not on the overall image thus will not be able to recognise it.

3. Encouraging autonomy

In terms of education, another important sphere of interest regards the autonomy of the child. Basic autonomies must be conquered step by step but education and therapies should begin rather early. In this regard, it is fundamental to reflect on our personal emotional experience as parents. For example, parents can develop a protective attitude that may lead them to undertake all the child's duties on their behalf. This is because a parent can be inwardly convinced that the child is too fragile and in need of help with everything. It will therefore be useful to be aware of these thoughts, and without denying it, try not to identify with them and keep in mind that certain autonomies can be taught and learned by the child. This renewed attitude on its own will surely make the child less fragile and needy.

- **Feeding habits.** As soon the child is able to eat solid food, this should be cut into small pieces and the child should be encouraged to eat with their hands. Tip: you can put a sheet of newspaper on the floor in order to collect what the child will inevitably throw on the floor. It is also advisable to put a few pieces of solid food at a time and place them directly on the tray of the high chair. One can begin gradually, firstly starting the meal by presenting some tasty treats, then proceeding by feeding the child the rest of the food and finally making the child eat everything on their own (always presenting a little food at a time, never in large quantities). Cutlery should be introduced later on when the child has learned to eat everything with their hands (see page 8).



- **Sleep.** The main aim is to get the child to sleep alone in their bed and secondly succeed in getting them to fall asleep on their own (see page 6).
- **Dressing up and undressing.** From the age of two we can ask the child to cooperate in these activities by offering them easy tasks at first and progressively more difficult ones (see page 7).
- **Sphincter control.** The majority of children with SCDC reach a good sphincter control even if the timing may be quite variable and the age at which to begin working on this may change from child to child.
- **Going to the bathroom.** Self-care can be taught by creating routines in which the child is personally accompanied by a parent at first, then verbally guided and finally left on their own.

4. Researching for and participating in rehabilitative programs

Both field experience and numerous research studies attest the importance of family participation in the child's educational and rehabilitation programs. A constructive form of participation must take an active approach rather than a passive one. An extreme situation is when the therapy is conducted behind closed doors and where largely oblivious parents assume the role of mere chauffeurs. Yet, nowadays in most cases therapists share with parents the aims and modalities of their work with the child. Nonetheless, therapists often have a merely informative attitude towards parents.

However, what really makes a difference is to propose activities that the parents will be able to do alone back at home. In order to learn, a certain repetition of the action is required. It is necessary to be exposed to the same experience several times in order to be able to internalise the process and its learning outcomes. This is why the best exercise done twice a week may not be sufficient.

There are also many practical questions to be taken into consideration: the eventualities of the therapist being on holiday, sick or busy with other important meetings may cause delays and hinder the therapy process. Similarly, the child may be ill or be unable to travel to the place of therapy, thus learning opportunities are further reduced. For all these reasons, and not because parents should become the therapist, parents are advised to share responsibility with the rehabilitation team, carrying out the rehabilitation in parallel with them.

5. Preventing and treating

The table below provides a summary of the possible medical problems and the corresponding treatments and information [cf. 5]:

CARE GUIDELINES		
PROBLEMS	CLINICAL DATA	DIRECTIONS
Neonatal problems	<ul style="list-style-type: none"> • Low birth weight, possible respiratory problems (asphyxia, cyanosis) • Difficulties in sucking, frequent vomiting, hypotonia 	<ul style="list-style-type: none"> • Neonatal and paediatric care • Psychological support for the family • Beginning the physiotherapy treatment from the first weeks of life (to enhance sucking and swallowing) • Possible breast feeding
Congenital malformations (rare)	<ul style="list-style-type: none"> • Cardiac malformations (septal defects, patent ductus arteriosus) • Intestinal malformations (mal rotations, Hirschsprung disease) • Cerebral malformations (agenesis of the corpus callosum) • Kidney malformations • Congenital dislocation of the hips • Cryptorchidism, inguinal hernia, syndactyly, cleft lip and palate 	<ul style="list-style-type: none"> • ECG, chest X-ray, echocardiography to diagnose BD • Abdominal ultrasound when it is deemed necessary • Ultrasonography after the diagnosis, CT and MRI if indicated • Urogenital ultrasound at 2 months • Ultrasonography of the hips at 2 months • Consulting a paediatric surgeon
Neurological disorders	<ul style="list-style-type: none"> • Potonia followed by hypertonia, psychomotor retardation and language delay • Sensorineural deafness (rare) • Convulsive seizures (rare) 	<ul style="list-style-type: none"> • Early rehabilitative interventions (from the first weeks of life): physiotherapy, psychomotor therapy, speech therapy. A close collaboration between the family and the professionals is fundamental • Audiometric test in the early months of life • Electroencephalography (EEG)
Anaesthetic problems	<ul style="list-style-type: none"> • Possible difficulty of intubation due to anomalies of the larynx 	<ul style="list-style-type: none"> • Inform the anaesthetist
Recurrent infections	<ul style="list-style-type: none"> • Respiratory and gastrointestinal infections 	<ul style="list-style-type: none"> • Immunological and allergological assessment • Compulsory and recommended vaccinations
Ophthalmologic disorders	<ul style="list-style-type: none"> • Divergent strabismus • myopia • cataract 	<ul style="list-style-type: none"> • Periodic Ophthalmologic evaluation
Orthopaedic disorders	<ul style="list-style-type: none"> • Flat feet • Talipes equinovarus • Scoliosis 	<ul style="list-style-type: none"> • Periodic orthopaedic checkup
Dental problems	<ul style="list-style-type: none"> • Presence of dental cavities (not too high) • Frequent malocclusion, such as an open bite 	<ul style="list-style-type: none"> • Early oral hygiene, fluoride, regular professional oral hygiene sessions • Orthodontic treatments, preferably under local anaesthesia
From: "Cri du chat syndrome" - Care Guidelines, Published by ABC Associazione Bambini Cri du Chat, San Casciano in Val di Pesa - Firenze, 2002.		



DIRECTIONS TO THE SCHOOL: PRINCIPLES AND PROCEDURES

The school environment can make it easier for the child to learn routines and rules of behaviour but at the same time it requires considerable effort from a sensory point of view. The school environment, since nursery, is usually full of noises in the foreground and background like screams, shouts, echoes, reverberation, etc... and very rich in visual stimuli such as colourful drawings, posters, backpacks, notebooks, etc... This is a huge inconvenience for a child with SCDC. After a few months the child may have learnt to tolerate and adapt to environmental noises, but probably they will not be able to listen, and consequently understand linguistic messages. This is because all their energies are invested in controlling the sound of the environment. Therefore, it is very important that the school takes into account of these difficulties and tries to organise a more facilitative environment for the child. In doing this a cooperation between the rehabilitation team and the parents is fundamental [cf. 23].

Some common issues are addressed below:

Does the child need to be in the classroom as much as possible or not?

Many teachers are concerned about trying to promote the socialisation of the child looking for their participation in classroom life and avoiding as much as possible keeping them apart from the other schoolmates.

In this regard, two observations must be made:

1) Socialisation is a process that must be gradually built up. It should start from the construction of a two person relationship (the child and the support teacher), which can then be expanded to a small group of children (3 or 4) through the mediation of the (support teacher). We should also consider socialisation to be a long term goal as the overall result of the educational process. Socialisation should not be imposed to the child as soon as they enter school, neither can it be seen as the premise from which to start. Indeed, directly entering the child in the whole class group can be for him or her an overloading sensory commitment. This will be especially true if the relationship with the support teacher is not yet structured and defined. This can increase a sense of unease and disorientation facilitating the re-emergence of stereotyped and problematic behaviours that may alienate the child from the other classmates.

2) The second consideration concerns the child's academic learning. It is highly demanding to request the child to understand and execute educational tasks in a context that in itself requires great effort and a huge sensory control.

Thus, in addition to moments where the child is integrated with the rest of the class, it is essential to build a personal relationship and work individually with the support teacher in a dedicated environment.

School: for what purpose?

The basic goals of going to school are learning to read, write and practise arith-



metics. Yet, when concerning a child with disabilities, other aims often become more important. These are such as socialisation, the pursue of autonomy and integration. Nonetheless, if we want these words not to be without meaning, the standard learning goals (reading, writing, arithmetic) must also be considered conceivable for these children. The school should be primarily oriented to the promotion of scholastic learning. For an SCDC child, reading, writing or computing skills may seem precluded. Surely the level of global development has to be assessed and sub-goals that aim to promote the pre-requisites for such skills should be set.

Reading involves a fundamental prerequisite: an adequate visual attention. A Cri du Chat child finds it very difficult to keep the focus on an object and prefers to observe moving items. Thus, in the process of learning to read, it is recommended that the child works on object recognition. Firstly this can be applied to pictures and finally to symbols such as those of the alphabet. At the same time, in learning how to read, another prerequisite is necessary: a sufficient auditory attention that can allow the child to listen attentively and understand. When a function is absent this can be decomposed into several sub-goals. It just needs to be clear how the function concerned is structured. This is exactly what should be done when teaching an SCDC child how to read: step by step, starting with sub-goals. Some children will only be able to learn how to recognise and deliver meanings with respect to objects, while others will be able to learn how to read even if just in a basic manner.

Regular school or Special school?

Another question that parents often face is whether they should enter their special needs child into a normal school or a special institution. A special school offers small classes to children with various types of disabilities and typically it will be the same institution as where the child undertakes the prescribed therapies. Inducting a child into a special institution may be simpler, more convenient and in some cases advisable. In general, however, experience suggests that a guided induction to a regular school is highly preferable. This can trigger a process of imitation and motivate the child with disabilities.

Handy tips to structure the environment

1) Create a comfortable study corner: the desk should be facing a blank wall and have on its side shelves containing the necessary materials.

2) Short Activities: the duration of an activity or a therapy depends on the attention span of the child. Nonetheless, the proposed activity should be suitable for the child to complete within a few minutes. Specifically, the expected time should never exceed 10 minutes after which the child will need a short break before starting again.

3) Plan ahead: It is important to have in mind the lineup of activities to offer and organise the materials you will need before starting the activity. In this way you avoid creating empty moments, which reduce and distract the child's attention.

4) The proposed activity must be completed by the child and it is worth emphasising the beginning and end of this activity.



- 5) Propose one activity at a time.
- 6) Speak quietly and calmly with no changes in the voice tone: whispering can also be useful to capture the child's attention and to help them to complete the presented tasks.
- 7) Messages and instructions must be simple and with few essential words to facilitate the child's understanding.
- 8) In some cases the attention span of the child can improve if the environment is in dim light (e.g. by placing a dark curtain on the window). In any case, the child's desk should not be near the window and should not have direct or reflected light upon it.
- 9) Set clear rules regarding the activities.
- 10) The images used must have limited visual details. For example it is better to propose an image with a single subject on a neutral background.
- 11) Due to the child's visual difficulties, we recommend the use of grey paper with no lines or squares. Avoid also recycled paper sheets, which have a dotted texture. In computer activities instead, it may be useful to reduce the brightness of the screen.
- 12) Music may be used in specific workshops and is usually a greatly appreciated experience. It is better not to play music during other activities. This may interfere with the child's listening and understanding of the vocal messages and instructions of the proposed activity.

GUIDELINES TO THE REHABILITATION PROJECT

The rehabilitative intervention should simultaneously move on the individual dimension of the child (promoting their highest possible potentials) and on the context and environment they are living in. Therefore, the family needs to be fully involved in the rehabilitation program coordinated by a specialist team, which may include paediatrician, neurologist, physiatrist, physical therapist, speech therapist, psychomotor therapist and occupational therapist. It is very important not only to inform the family on which therapies the rehabilitation project includes, but to consider its fundamental role in the evolution of the child and thus directly involve the family in the rehabilitation project.

The most positive results are recorded in correspondence of a comprehensive therapy. Comprehensive therapies are a type of treatment that consider the child in their specific stage of development, their personal resources/skills, their deficits and their family environment. Physiotherapy, speech and psychomotor therapy will therefore not be sectoral (i.e. focused on a single aspect) but transversal, trying thus to take into consideration the prerequisites that underpin the development of several functions.

Thus, in the creation of an individual rehabilitation project many levels overlap:

INDIVIDUAL DIMENSION

- REHABILITATION OF SENSORY DISORDERS
- MOTOR DEVELOPMENT



- DEVELOPMENT OF LANGUAGE AND COMMUNICATION SKILLS
- DEVELOPMENT OF AUTONOMY
- CONTROL AND MANAGEMENT OF THE BEHAVIOUR
- DEVELOPMENT OF THE ATTENTION SPAN
- DEVELOPMENT OF THE CAPACITY TO COMPLETE WORKS AND ACTIVITIES
- DEVELOPMENT OF SYMBOLIC AND COGNITIVE CAPACITY
- DEVELOPMENT OF ORAL MOTOR FUNCTIONS
- DEVELOPMENT OF MANUAL FUNCTIONS

CONTEXT DIMENSION

- INFORM THE FAMILY
- SUPPORT THE FAMILY
- INVOLVE THE FAMILY AND ASK FOR ITS PARTICIPATION IN THE EXECUTION OF THE CHILD'S ACTIVITIES AND THERAPIES

Nonetheless, the existence of many possible levels of action must not create a chaotic pot where everything is thrown in. Sometimes programs are very articulate, full of goals and proposals but often result fragmented and difficult to verify. It is instead important to highlight priorities, resources and deficit areas and it is fundamental to understand which are the prerequisites underpinning the development of one or more functions that are not yet developed. Thus, the role of the rehabilitation specialist is primarily that of a careful evaluator able to observe the child in their overall performances and characteristics and thus indicate to parents and school staff the most suitable activities, the modalities and the objectives to be pursued.

Moreover, it is important to have a single rehabilitation program with detailed activities and few clear and verifiable objectives. The program needs to be carried out by the parents at home, by therapists in the specific rehabilitation institutions and by teachers at school.

In the appendix you can find an empirical example of a Cri du Chat girl. It outlines a comprehensive evaluation of her condition, the rehabilitation project undertaken and the re-evaluation after one year of therapies.



EVALUATION OF THE CHANGES AND DEVELOPMENTS INDUCED BY THERAPIES IN A SAMPLE OF THIRTY ITALIAN CHILDREN AND TEENAGERS AFFECTED BY THE CRI DU CHAT SYNDROME: MAIN FACTORS

METHOD

Over the years 2004, 2005 and 2006 thirty Italian children and teenagers affected by SCDC have been under observation in order to produce this study. The aim of the study was to produce a comprehensive overview of the behaviour of the children and teenagers. The study included the use of some tables of evaluation and a parallel observation of the spontaneous behaviour of the children. Here we took into consideration the development of linguistic skills based on some data extracted from the MacArthur Questionnaire [cf. 2]. The final goal was to understand as objectively as possible how these children and teenagers improved their language and communication skills during the course of one year of therapies.

The Mac Arthur questionnaire can be used to assess communication and lin-



guistic skills during the first year of life or in children who are at a stage of development inferior to their chronological age.

Since the age of the thirty children and teenagers who participated in the study was very broad (from 1 year to 18 years), it was decided to divide them into 3 age groups: - 1 to 4 years, - 5 to 9 years and - 9 years and over.

We chose not to use IQ and mental age assessment tests, as we judge them to be unreliable and poorly significant in regards to the actual mental ability of children affected by SCDC. In fact, characteristics of this syndrome are an attention deficit, sensory alterations and behavioural difficulties. These aspects do not allow an easy distinction between what are cognitive difficulties and sensory and behavioural ones. Instead, we believe the MacArthur Questionnaire to be significantly more indicative, as it is based on the parents' observation of spontaneous behaviour acted out by the child. In our study, based on this test, an outside therapist was substituted to the parents in the role of the observer. The test, which is set on records of the non disabled population, reports the number of words that should be understood at 17 months and produced within the first 30 months of life.

The test consists of 2 tables: the first table includes a list of 230 nouns, 55 verbs and 63 gestures and the second table consists of a list of those 230 nouns plus another 133 nouns (tot.363), the same 55 plus another 48 verbs (total 103) and 104 elements related to phrasal organisation (e.g. use of paired words, use of simple sentences subject-verb-object, use of free morphological elements (articles, prepositions, clitics or related decline singular-plural, subject-verb agreement)).

RESULTS OF THE FIRST EVALUATION

Group 1: 10 children aged between 1 and 4 years.

- 60% of the children understood more than 30 nouns (e.g. mummy, daddy, hunger, yes, no, bed, etc.)
- 50% of the children understood more than 5 verbs (e.g. eat, sleep etc.)
- 50% of the children used more than 10 intentional communicative gestures (e.g. pointed at an item to ask for it, answered yes / no with their head, etc.)
- 40% of the children were able to say at least one word.

In general it was noted that, if a child understood many words, he or she will produce even more gestures of communication to request something and connect with others.

Nonetheless, a high variability between each child was highlighted and this is partly connected to the type and severity of the genetic deletion.



Group 2: 10 children aged between 5 years and 9 years

- 60% of children understood more than 100 nouns
 - 50% of children understood more than 20 verbs
 - 80% of the children used more than 20 communicative gestures
 - 60% produced at least 18 nouns (of which 30% of children use over 150 names)
 - 30% of children reproduced more than 10 verbs (of which 20% use over 50 verbs)
 - 40% paired two or three words (e.g. mom - ball)
- Yet, phrases are not structured.

However, it was also noted that in 3 out of the 10 children observed, the expressive-verbal language was absent.

Specifically, it was discovered that 2 of these children showed great difficulty in understanding words and used few or no communicative gestures at all.

However, one child showed a high understanding, a good use of gestures, but the expressive-verbal language was still absent.

Group 3: 10 children older than 9 years

- 70% of children understood more than 40 nouns (and 40% of them more than 300 names and more than 100 verbs)
- 50% of children reproduced more than 10 nouns (and 40% reproduced more than 100 names and more than 50 verbs)
- 40% of children could structure a sentence (in a more or less complex manner)

With regard to the 4 children with the highest levels of expressive language, we could affirm that the instrument used for the evaluation was not suitable. This is because their vocabulary, both in the understanding and in the reproduction of nouns and verbs, was much more extensive (children are using more words than those considered by the MacArthur Questionnaire) and the number of these words is no longer measurable with ease.

However, we also noted that in 5 out of 10 children the expressive language was absent or corresponded to less than 4 words. Of these children, 4 out of 5 had very little or no understanding and were unable to use gestures for communication. Just one child showed instead gestural communication skills through which they were able to communicate some of their needs (the use of facilitated communication was introduced with good results but only in the presence of the facilitator).

THE FIRST EVALUATION: SOME OBSERVATIONS

- 1) We observed that communication skills and the ability to understand verbal



language develop **within the 4th year of life.**

At this stage the reproduction of words is still very poor and the preferred communication appears to be non-verbal gestures. Detecting the presence of communication gestures is very important because this indicates that the child has understood their symbolic value. The child understands that an action causes a reaction (e.g. the child points to the glass of water so that the mother brings him the water). Consequently, the child can develop an active relationship with the environment and this is indeed the underpinning mechanism at the core of verbal communication. In fact, verbal language is just an extension and evolution of this process. Therefore, we can affirm that those children who use many gestures within their first 4 years, will most likely develop verbal language skills later on.

2) **From 5 to 9 years**, a development of the child's vocabulary and an increased understanding and use of words is noticeable. However, the phrase is not yet structured. The use of paired words or extremely simple phrases was also noted in some cases (e.g. "Mum - food" used to express their hunger).

3) It is **after 9 years** that verbs become widely used in verbal communication. Moreover, the appearance of elementary sentences is observed (Subject-Verb-Object) with elements of free and bound morphemes (articles, prepositions, etc...). Whilst the child develops the use of sentences, they continue to expand their vocabulary.

4) With regard to the observations concerning the **lack of verbal communication** we did not consider the first group of children (under the age of 4 years) because their expressive verbal skills at that age are not yet structured. Observing the other 20 children, instead, **it was discovered that 8 out of 20 children and teenagers did not possess verbal skills**. Of these, 6 children also had very basic non-verbal communication skills (such as the use of gestures to ask for things). It was also noticed that these 6 children had a very limited ability to relate with their surrounding environment. On the contrary, the other 2 children showed a good understanding and discrete use of non-verbal communication skills (use of gestures and of Facilitated Communication).

This appears only partially linked to the genetic deletion. In fact, just some of these children had more severe deletions, while in many others the deletion had a medium level of severity.

In particular, looking at children within the same age group, we noted that even children with the same chromosomal breakpoint (thus with similar genetic alterations) showed completely different development profiles.

For example, in a Cri du Chat child we observed that the language development was completely absent and that the gestures of communication were very limited and not spontaneously used. At the same time, abnormal behaviour, sensory deficits and reduced development of basic autonomy were reported. In contrast to this example, another child with the same chromosomal break point showed rather good verbal language skills (i.e. a high number of words used and basic phrasal organisation). Furthermore, the child's behavioural difficulties were far more manageable and he showed a good development of basic autonomy. We have also observed examples



of children with severe genetic deletions who demonstrated a more advanced level of development than other children with minor genetic deletions.

Variability is thus very high and the genetic deletions do not appear to be the only determining factor in the overall development and more specifically in a linguistic sense.

5) By expanding the evaluation to the other 12 children who showed evolving language skills, we were also able to make some considerations in relation to other aspects of language development that are not investigated by the MacArthur Questionnaire.

In particular we noted the presence of various articulatory, phonological and morphosyntactic limitations. With regard to the phonological level (i.e. the level of the sounds) we observed that some of the speech sounds were absent: for example children could not pronounce the sounds “r”, “s” and certain types of softer sounding “c” and “g”. Consequently, all the words that contain these sounds were altered and not always comprehensible. For example, if the sound “s” is missing from the word “music”, this can become “muki-k.” In addition to these missing sounds, we also observed many processes through which the child simplified the phonological structure of words. The more numerous the missing sounds and processes of simplification are, the more the child’s language is difficult to understand, especially for an outside listener who is not used to it. Moreover, children often showed articulatory difficulties that affect the production of sounds. In fact, these children reported difficulties in moving the muscles of the tongue, lips and cheeks to produce various linguistic sounds (apraxia). Other difficulties were instead related to malformations of the phonatory organs.

At a morphosyntactic level 3 out of 12 children were observed as being able to organise basic sentences using also the main morphological elements (prepositions, articles, coordination between the gender and number of nouns and verbs etc...). In 9 out of 12 children, instead, the phrase was limited to a maximum of 3 words and more complex morphosyntactic elements were absent. In these cases, verbs were not used and the sentence was actually made up of 2-3 ordered words. Finally, one last consideration on issues of language development. For the appropriateness of language development we mean a number of things: the communicative content language development, its appropriateness to the context, the ability to modulate verbal behaviour in different situations and the use of irony, etc.... If we consider the 6 children with the highest levels in the MacArthur questionnaire, only 1 of these showed a good level of appropriateness in language development. The other children, at different levels, instead showed reduced competence: verbal language seems redundant and poor in terms of content and often not adequately or sufficiently modulated according to the context. Nonetheless, in all these cases, speech therapy is highly recommended. In particular, we emphasise that even for children with no expressive language skills, speech therapy may be beneficial. This is especially true as long as the therapy focuses on the development of the prerequisites of verbal language skills and works on the behavioural and sensory abnormalities that underlie the children’s language difficulties. In this case the therapy is not a speech therapy in a strict sense but rather a therapy with a more comprehensive character. Therefore, the direct involvement of parents and teachers in the rehabilitation program is fundamental.



RESULTS OF THE SECOND EVALUATION
AFTER A YEAR OF COMPREHENSIVE THERAPIES
(not necessarily focused on language development)

Group 1: the age of the 10 children ranged from 2 to 5 years

Understanding:

70% of the children understood more than 40 nouns and 50% of them could understand more than 100. In addition, 60% of the children could understand more than 15 verbs and 30% of them more than 30.

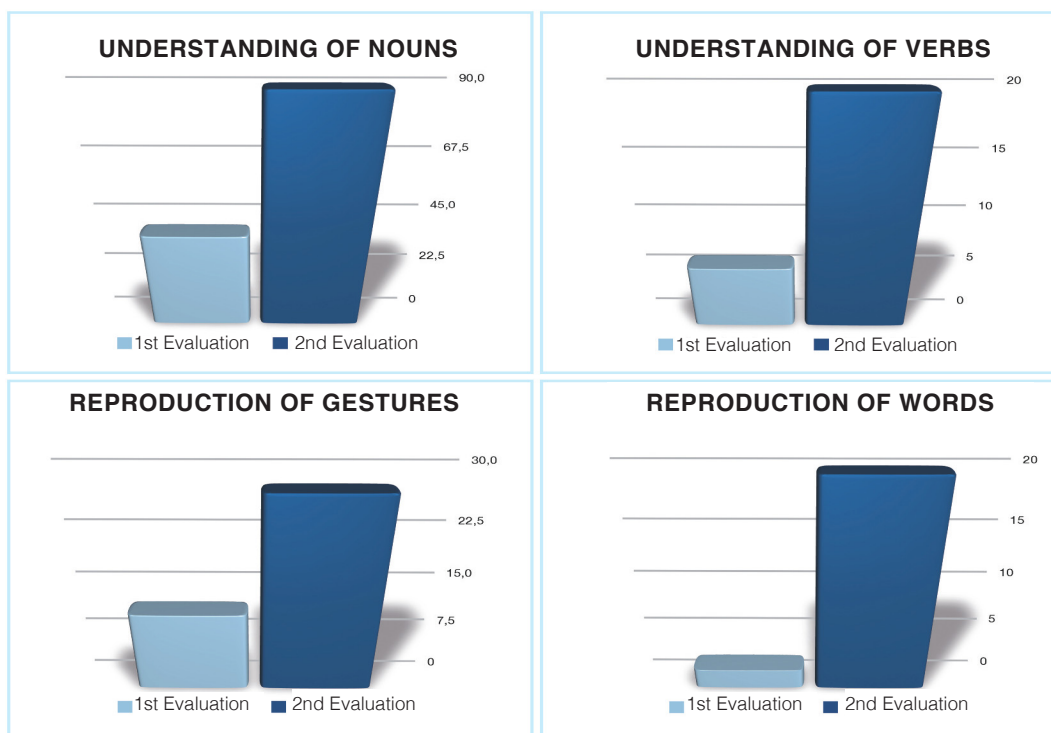
Gestures:

60% of the children reproduced more than 20 gestures.

Words reproduced:

80% of children used at least 2 words and 20% of them were using more than 50. Gestural communication remains the most frequently used mode to express their needs.

An average - differences between the first and the second evaluation in the 1st group of children:



Note: in all charts, the maximum value is related to the number of elements considered by the test, thus the value of the average is a value in reference to the test.



Group 2: the age of the children ranged between 6 and 10 years

Understanding:

- 80% of the children understood more than 100 nouns,
- 70% of the children understood more than 20 verbs

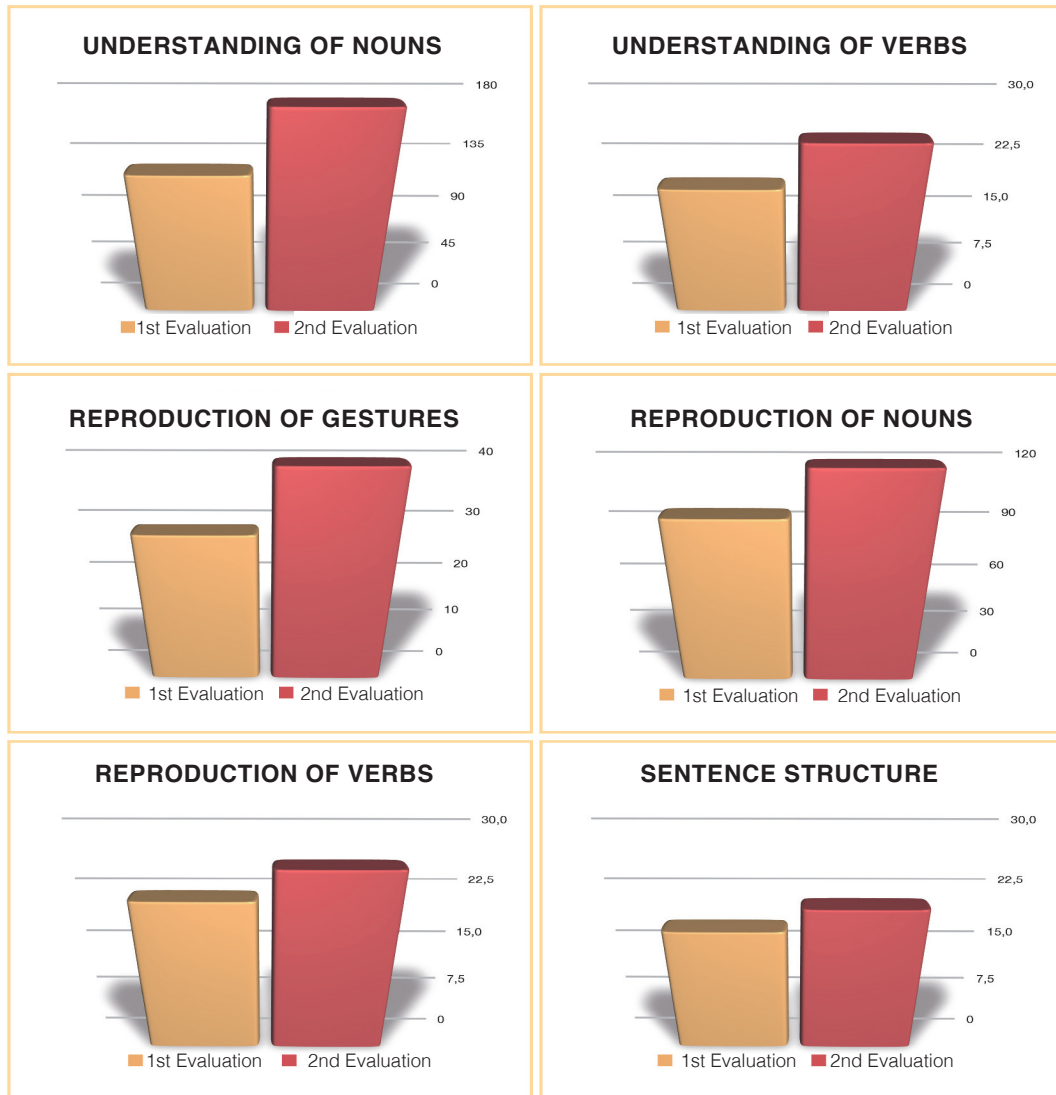
Reproduction of gestures:

- 80% of the children use more than 20 gestures

Reproduction of words:

- 70% of the children used more than 18 nouns and of them 50% were using more than 150
- 50% of the children used more than 10 verbs
- 50% of the children used very simple sentences (2-3 words), with few morphological elements

An average - differences between the first and the second evaluation in the 2nd group of children:



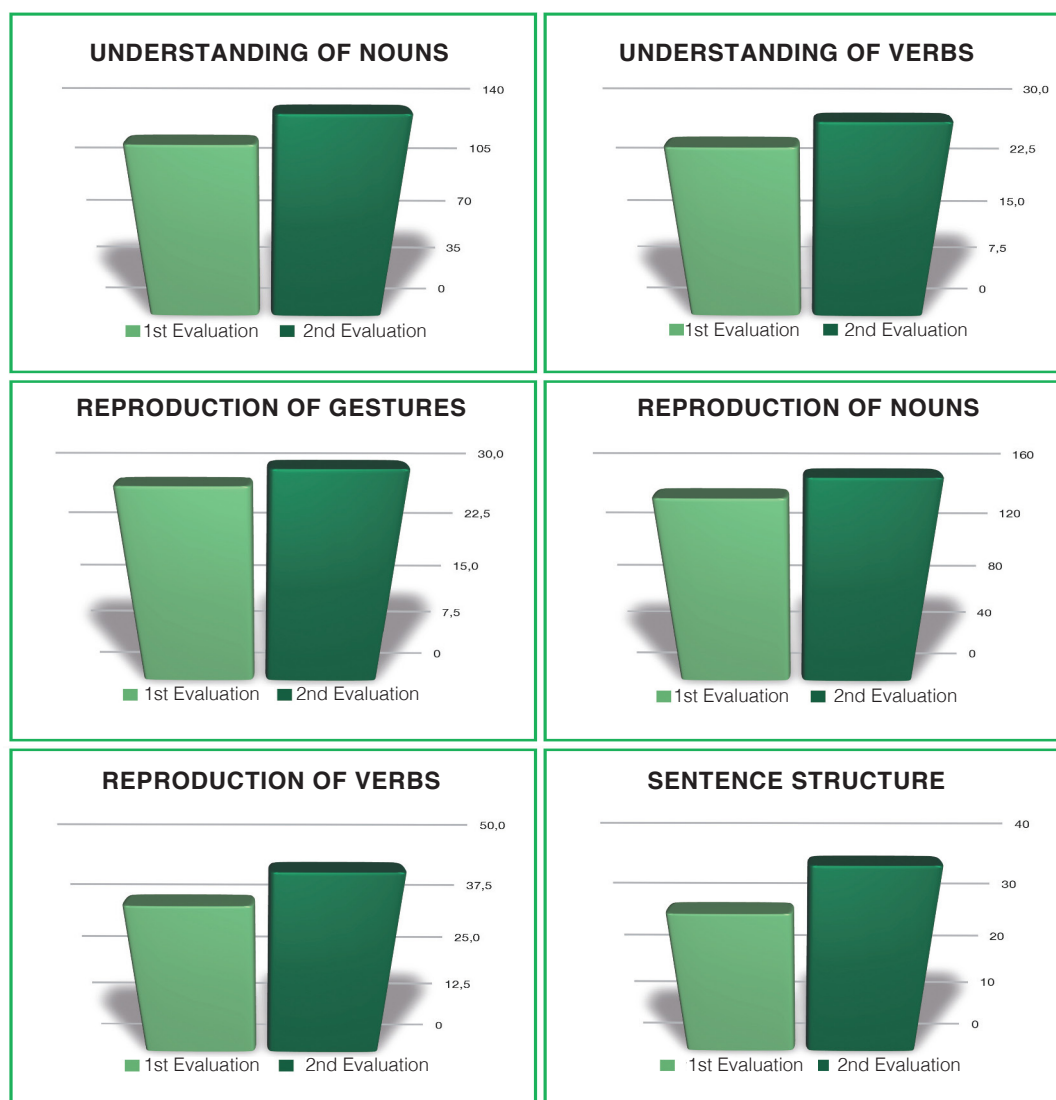


Group 3: Youth's from 11 years

- 70% of the teenagers understood more than 40 names and 40% of them could understand more than 300 names and over 100 verbs
- 60% of the teenagers reproduced more than 10 names and 40% of them could reproduce more than 100 names and more than 50 verbs

In 40% of the teenagers the sentence was organised (in a more or less complex manner)

An average - differences between the first and the second evaluation in the 3rd group of children:





DATA INTERPRETATION

Group 1: The improvement of the children is evident in all areas (a bit less in terms of word reproduction). Observing each individual situation it can be noticed that only one child out of 10 does not show significant improvements, while all the others, albeit with great variability, show important changes especially in the sphere of language understanding and use of intentional communication gestures (e.g. responding yes / no through gestures, indicating an object that they want etc ...) . In addition, an increased capacity of expression and quantity of words used is noted.

Group 2: Even within this group the improvement of the children is evident in all areas. Just the linguistic - communicative skills of two children remained almost unchanged. All the other 8 children showed instead both an increase in their understanding and verbal communication. In particular it was observed an increase in number of words used and a greater use of morphological elements, even if the sentence on average remains very simple.

Group 3: From the charts we can observe that, compared to the charts of the first two groups, the improvement of these teenagers was much more modest across all the different areas. This was for two simple reasons. Not just that for 4 children out of 10 the MacArthur Questionnaire reached its plateau during the second evaluation , but it already did so during the first evaluation. Thus, important changes are not shown in the charts even though the language skills of the Cri du Chat teenagers continued to increase. As previously noted, the questionnaire is not a suitable instrument to assess language development in this case. The language skills of this group of teenagers was, in fact, more developed than the group investigated by the test.

The other 5 children and teenagers who at the first evaluation had practically no language skills and extremely poor communication skills showed very limited changes after one year. In three children, specifically, the situation remained unchanged. Two children showed a greater use of some gestures of communication, some improvements in the understanding of some verbal messages and the ability to use 3-4 words.

Observing the children with the greatest difficulties of communication, we also noted that they showed marked sensory difficulties at various levels (see pages 10-13) and that they were often “trapped” in a series of stereotyped behaviours, which contributed to severely limiting their ability to create a relationship with the external environment.

In parallel, children and teenagers with more or less structured language skills present less problematic and stereotyped behaviour, more autonomy and a greater learning ability. We can therefore hypothesise that the sensory difficulties at the foundation of behavioural problems, also prevent the understanding of the communicative value of gestures and words and hinder the children’s attention to verbal language.



It should also be noted that the sensory aspects are not covered by any specific rehabilitation treatment. Instead, sensory difficulties are often classified as general disorders and therefore remain untouched by the rehabilitation programs of physiotherapy, psychomotor and speech therapy that are typically activated immediately.

Another consideration that can be drawn has to do with the family and school environment in which these children are grown. In fact, the 30 children on who we tested the MacArthur Questionnaire were evaluated within their everyday standard environments, both at home and at school. The internal dynamics and ways of managing the child and the related difficulties were also observed. Without going into the specific details of each situation, we can affirm that the attitude of the people who follow the child in his or her education is a very important factor in the child's development. Thus, special consideration should be given not just to the therapies themselves, but equally to the specific environments and manner upon which this is continued. By environment we are referring to the family system and the school environment where the child is spending most of his or her time. From this point of view, an attentive and caring attitude marked at the same time by setting clear rules and clear teaching can be very good for the child to organise their behaviour in a constructive way.

Moreover, even the collaboration and participation of parents and teachers in the education projects is fundamental to convey positive stimuli for the child's behaviour. This is a very significant aspect since the behavioural disorder, characterised by hyperactivity and problematic behaviours (resulting from sensory deficits and self-regulation deficits, including in some cases the organisation of the sleeping rhythm) forms the "background" of the child with SCDC and hinders or slows down their learning and cognitive development. When the environment is organised in rituals and rhythms, the child always knows where he is, and what may happen to him or her. They are reassured and thus helped to organise their own behaviour. In this way a Cri du Chat child will be able to use their resources primarily to relate with the environment and consequently to learn.

On the contrary, when landmarks are unclear, rules are ambiguous, constantly negotiated and the overall management is fragmented and the child is not facilitated by the environment a Cri du Chat child will present greater behavioural difficulties. These difficulties, which can be very hard to manage, will ultimately distract the child from the relationship with the external environment and the learning opportunities presented to them.

Effective rehabilitation projects have been aimed at directly involving the parents, giving them support, information and programs to work with the child on their own. They have also promoted contacts between parents, they have tried to replace the dysfunctional strategies and behaviours spontaneously adopted by parents with more functional approaches and have shared with them the principles and techniques of the therapies.

The type of therapies proposed and the precocity of the rehabilitative intervention are also key considerations. Therapies can begin almost immediately through the



support of the family and the presence of specific guidelines. It is not necessary for the child to do physiotherapy three times a week if the physiotherapist provides the parent with the necessary information. This is not because the parent has to become a therapist, but because parents are in contact with their child all the time and not just during the hours of the therapy session. Therefore, it is fundamental to provide the parents with the means to actively stimulate their child in their educational programmes.

In the rehabilitation program it is also fundamental to look at the causes of the symptoms before beginning to work on the latter. For example, the absence of language is a symptom and the related potential causes should therefore be analysed. Let us take a concrete example: in the case of a child of 6 years with stereotypes, sensory and behavioural problems, who is unable to communicate and even has difficulty in the process of understanding verbal communication, the speech therapist will have little success if the therapy will aim just at setting samples of sounds to be articulated by the child. On the contrary, a therapy that seeks to organise the sensorial capabilities of the child and promote their auditory attention to language will achieve better results.

















Another important aspect of the rehabilitation program is its precocity. In more recent years there has been an overall high level of precocity in the early stages of therapies, while in the past it was very common to start rehabilitation treatments almost too late. Young children appear to be facilitated by early and prompt therapies that act not just on the child, but also on the parents by providing them with the tools necessary for a very difficult task: the education of the child.

Considering the above observations it follows that, beyond the aspects of genetic and genotype-phenotype, other aspects related to family and school environment and quality of rehabilitative interventions seem to have a very high and significant incidence in the promotion of psychomotor and language development.



APPENDIX 1

Some useful advice, not in order of importance

-  RULES MUST BE CLEAR AND MOTHER, FATHER AND GRANDPARENTS SHOULD ALL AGREE ON THEM, ESPECIALLY IN THE CONTINGENT MOMENT
-  FIX A FEW RULES AT A TIME, WHICH MUST REMAIN CLEAR AND RESPECTED IGNORE THE CHILD IF, TO OBTAIN ATTENTION, THEY BANG THEIR HEAD OR BITE THEMSELVES etc ...
-  DO NOT ALLOW THE CHILD TO BITE YOU, PINCH YOU OR PULL YOUR HAIR
-  NEVER REPROACH THE CHILD WITH A SMILE
-  A “NO” IS FINAL AND NOT NEGOTIABLE
-  NEVER SUCCUMB TO “BLACKMAIL”
-  ORGANISE THE DAILY SCHEDULE OF THE CHILD THROUGH REGULAR RHYTHMS
-  CREATE ROUTINES, HABITS AND RITUALS
-  EDUCATE THE CHILD NOT TO DO THINGS THAT WILL NOT BE APPROPRIATE WHEN HE OR SHE IS GROWN UP
-  BEWARE OF TOO MUCH BACKGROUND NOISE - CREATE A QUIET CORNER IN WHICH YOU PROPOSE ACTIVITIES TO THE CHILD
-  SPEAK QUIETLY ESPECIALLY IF YOU WANT TO ATTRACT THE ATTENTION OF THE CHILD
-  SPEAK TO THE CHILD WITH A CALM AND CONSTANT VOICE
-  SPEAK TO THE CHILD WITH SIMPLE AND CLEAR MESSAGES (USE FEW WORDS!)
-  SEARCH FOR EYE CONTACT WITH THE CHILD
-  ONE GAME OR ACTIVITY AT A TIME
-  AVOID NOISY AND CHAOTIC PLACES



- 👁️ TOO MANY PEOPLE, STIMULI OR GAMES SIMULTANEOUSLY WILL DISTRACT THE CHILD
- 👁️ THE CHILD MUST SLEEP IN THEIR OWN BED!
- LET THE CHILD EAT ALONE, WITH THEIR HANDS AT FIRST AND WITH CUTLERY AFTERWARDS
- 👁️ TRY TO MAKE THE CHILD EAT, SLEEP, GO TO THE TOILET AND DRESS AND UNDESS THEMSELVES ALONE WITH AS LITTLE HELP AS POSSIBLE
- 👁️ DO NOT DRESS THE CHILD TOO MUCH AND LET THEM STAY BARE FEET INSIDE THE HOUSE
- 👁️ ASK FOR HELP AND SUPPORT
- 👁️ SEARCH FOR A DIALOGUE WITH TEACHERS, THERAPISTS AND PHYSICIANS
- 👁️ TALK WITH PARENTS WHO ARE EXPERIENCING SIMILAR DIFFICULTIES
- 👁️ CONSULT THE THERAPIST FOR SPECIFIC EDUCATION GUIDELINES
- 👁️ TASKS AND ACTIVITIES SHOULD BE BRIEF (A FEW MINUTES) BUT REPEATED MORE TIMES THROUGHOUT THE DAY
- 👁️ ALWAYS EMPHASISE TO THE CHILD THE BEGINNING AND THE END OF AN ACTIVITY
- 👁️ DO NOT BE IN A HURRY TO SEE THE CHILD WALKING ON THEIR OWN
- 👁️ TAKE CARE OF THE CHILD'S ORAL HYGIENE
- 👁️ DO NOT WORRY TOO MUCH IF THE CHILD DOES NOT EAT MUCH DURING THEIR EARLY YEARS BECAUSE THEY WILL EVENTUALLY CATCH UP!



APPENDIX 2

A case report: Anna, two years

Main notes on her medical history

Anna was born on time, her weight was normal, however there were problems with feeding and breathing. The diagnosis was made immediately after birth, confirmed by laboratory tests in which it was proven that a deletion of chromosome n° 5 of medium severity occurred .

Divergent strabismus without visual changes was discovered and auditory examinations resulted normal.

FIRST ASSESSMENT - SENSORY ASPECTS

Visual aspects: Eye contact is evasive, she recognises very minute visual details, her attention is drawn to wires and hair and she often moves her hands in front of her eyes, she swings objects, and she is attracted by lights and colours. Additionally, her gaze follows a moving object for a very short time, visual convergence is absent and she cannot distinguish simple visual symbols. Hypersensitivity.

Auditory aspects: Anna is scared by sudden loud noises and she tends to isolate herself when in noisy environments. She is sensitive and very attentive to whispers and low level sounds. When asked to, she performs simple tasks (especially picking up objects) and she shows an understanding of significant sounds. Hypersensitivity.

Touch: At a deep level her pain threshold is high (e.g. so that a fall may result less painful than a nip). Occasionally she pinches herself, hits herself, hits her head, especially when she is reproached. She cannot organise her stereognosis skills. She presents tactile hypersensitivity especially in her legs. Reduced deep tactile sensitivity, high tactile sensitivity on the skin's surface.

Motor skills: Anna begun rolling at 11 months and crawling at 13 months. She currently moves in cross pattern crawling but only for short distances and some months ago she acquired an antigravity axial control. She has also just begun to pull herself into a standing position by grabbing fixed objects.

Language: Anna does not show any significant production of finalised sounds. She reproduces whining and vocalisations. Only the sounds "t" and "d" are pronounced correctly. The crying is present but not loud. Occasionally there are some oral praxis of imitation. She does not use communicative gestures (to request or to point at) and she cannot grab items if she is requested to.

Manual skills: Anna grabs and drops objects but she does not oppose her thumb and index finger. Still she cannot perform finalised (and symbolic) actions with items because she appears too distracted by the sensory aspects of the items (especially tactile and visual).

Feeding and oral-motor aspects: Anna is rarely hungry and she is unable to chew and refuse solid food even if presented to her in small pieces. Swallowing is normal and she does not show any unusual reactions to smells. Sometimes, especially when ill, she refuses food and will vomit if presented with food. As an overall she demonstrates a very



irregular relationship with food. Anna presents also hypotonia of the lip muscles, the tongue is occasionally protruded and she drools.

Observation of spontaneous behaviour: Anna manipulates and slams objects, she licks and puts things near or in her mouths. She demonstrates a predominantly oral knowledge (i.e. to understand how things work she tends to put them first in her mouth) and she does not use communicative gestures. She does not show particular interest in exploring the environment and tends to remain very still. She does not communicate her needs. She is not noisy and her spontaneous vocal production is quite poor.

Problematic behaviours: Anna pulls off the glasses of people who get close to her, she licks objects, the floor and furniture. Anna also bangs her head and teeth on hard surfaces, especially if something is prohibited to her or she is reproached.

Autonomy: She eats some food alone and she sleeps alone, but she falls asleep only in the presence of her mother.

Sleeping at night: For Anna it is very difficult and the parents frequently use the syrup Noprom to facilitate the onset of sleep. She falls asleep with her mother by her side but sleeps alone in her bed once her mother leaves.

Ongoing therapies: Anna is currently doing twice weekly sessions of physiotherapy. *At an overall neurological level, Anna shows a neurological age between 7 and 12 months.*

THERAPIES PROPOSED TO THE FAMILY AND SUPPORTED BY THE CHILD PSYCHIATRIST AND THE PHYSIOTHERAPIST

Here the goals and activities proposed for a year of therapy are summarised (directions were updated every 4 months).

General instructions:

- Speak softly and whisper to attract her attention.
- Use facial expressions consistent with the verbal message (if reproaching her, keep an angry face)
- Do not approve attitudes that in perspective will not be appropriate (e.g. kissing everyone, clapping her hands for self gratification). In the presence of these behaviours, just let her be and do not emphasise them or avoid giving her too much attention.
- A '**NO**' must be clear, with a firm tone of voice, a few words and an angry expression.
- Leave her eating alone with her hands as much as possible.

ACTIVITIES PROPOSED WITHIN THE FAMILY FRAMEWORK

Duration: - around a minute for each activity
Frequency: - everyday
Intensity: - changes according to the different exercises
however activities should be repeated more than once throughout the day



1-Auditory aspects

Auditory attention to language: lulling whispering, whispering of onomatopoeic sounds, whispering of a nursery rhyme by modulating the tone of voice (e.g. angry-happy tone) or change the pace of the rhythm (spelling slower / faster). "Patting" on the child's head and ears.

2-Visual aspects

Visual attention: moving a light along the walls of a darkened room (for visual tracking). Stimulation with a red light at the corners of the eyes.

3-Touch

Body Massage: gentle touches , touches with a soft little brush and vibrating, rubbing and kneading soft massages.

4-Motor skills, balance and coordination

Crawling, increase the opportunities for crawling by making more space in the room (move the furniture in the room where the child spends more time), so that the child will have fewer opportunities to pull herself up.

Crawling up the stairs: useful for promoting visual convergence

Rocking: right-left and front-back.

» *Therapies on sensory aspects continue with an update on the directions given above on visual aspects, auditory aspects and touch.*

Autonomy

Let her eat a part of the meal alone or with her hands. (this means working on her autonomy, visual convergence and her fine manual grip)

Seek her collaboration in the process of dressing-undressing.

Behavioural strategies (also useful to give meaning to communicative gestures that per se are not communicative)

Let her pick up what she has thrown on the floor (this means working on the concept of cause-effect relationship and of the consequence of her actions). Ignore her and do not give importance to her provocations.

Try to respect what she wants. This means two things:

1) For example: when she shows that she does not want any more food, even if she has eaten little, respect her request and do not insist to feed her more.

2) If at the table she throws her glass on the floor, either let her collect it, or ignore her gesture, do not pick her glass up and do not give her more water during that meal.

The rejection must be clear and decisive, accompanied by appropriate facial expressions (do not smile when you want to blame her!). Is also very important to share this strategy: what the mother decide to do in a specific situation must also be supported by the other members of the family.

Attention to language and touch aspects (stereognosis)

- The game of taking the right item (e.g. between two visible objects, such as a ball and a doll, ask her to take the ball).



- Stereognosis games (e.g. put inside a cloth bag a set of items, such as a tennis ball and a book, and again ask her to pick one of them) .
- A useful everyday suggestion is for the adult to indicate an object as he/she names it.

SECOND EVALUATION: AGE 2 YEARS AND 11 MONTHS

Language and communication: Anna demonstrates improved non-verbal communication skills, interaction and eye contact. Since 2 months ago non-verbal gestures to request (she indicates the things that she wants) have appeared and the use of them is increasing. She can also answer yes / no through gestures. She shows and gives items to the adult. Five words are present in her verbal communication but in an erratic manner and she also performs a greater quantity of vocalisations. Anna still prefers a gestural mode of communication. Her verbal articulation is still poor. She has begun symbolic games linked to family routines, e.g. she feeds her doll. She shows good understanding and execution of orders related to the context (e.g she turns on the light, closes the door, opens her mouth etc... when requested to do so). She clearly understands the meaning of a "NO", but she does not always respect it. Even if not in a constant manner, Anna also manages to pick and give the right object to the adult when requested to select between two different objects. She has developed some oral praxis, such as blowing and giving a kiss. Her attention to vocalisation and to the language has improved, auditory hypersensitivity remains, she is still attracted by whispers and she tends to be restless in noisy environments.

Behaviour: Problematic behaviours have decreased. Anna bangs her head very rarely and only if contradicted. She slaps and pinches just in times of distress. She continues, albeit to a lesser extent, to orally explore objects (especially those objects that she has known for a long time). She has begun to play symbolic games even if often guided by an adult (e.g. playing with the doll). The use of the items is more functional albeit in reduced occasion she keeps using items to receive sensory stimuli (e.g. hit a little bottle of water on the wall to produce noise rather than using it to drink the water inside). She is definitely more present within her environment, she is more attracted and intrigued by what she sees and her spontaneous exploration of the environment has greatly increased.

Problematic behaviours: Sometimes she still pinches and gently slaps in an often provocative manner. When she is nervous she bangs her head backwards, but significantly less than before. Sometimes she throws objects on the floor to provoke the adult or to research sensory stimuli.

Motility: Anna is generally well organised in this aspect. She currently moves through crawling, which has become fast and effective. She crawls up and down the stairs in a well controlled manner. She shows a good side walk, she walks when supported by an adult with both hands, or when she pushes a trolley. The balance



is still poor. She grabs and drops objects, even little ones and she opposes thumb-index in one hand at a time.

Feeding: She presents the tongue-palate movement and starts to vertically orientate her chewing. She eats different foods and textures without major problems. She still uses a babies bottle in the morning. Also she drinks alone bringing the glass to her mouth with both hands.

Autonomy: Anna is mostly fed by her parents but she always starts eating the meal alone with her hands. She also collaborates in the dressing and undressing process.

Visual aspects: Anna follows moving objects in a non-continuous manner. She also recognises small visual details and she is still attracted to threads, hair and small crumbs. Sometimes she dangles small objects in front of her eyes. She manages to maintain a direct eye contact with the other party or with an object for a short period. She can follow and enjoy watching some cartoons that are visually simple.

Tactile aspects: Anna still shows hypersensitivity, especially on the sole of her foot. Her night sleep has improved and it is easier to comfort her when she wakes up during the night. Moreover, she falls asleep more often alone, especially in the afternoon.

The overall neurological age that Anna shows is between 12 and 18 months.



REFERENCES

1. ABC, Associazione Bambini Cri du Chat, "La sindrome del Cri du Chat, A.B.C. Associazione Bambini Cri du Chat.", S.Casciano in Val di Pesa, 2002.
2. Caselli MC, Casadio P, "Guida all'uso del questionario Mac Arthur per la valutazione della comunicazione e del linguaggio nei primi anni di vita.", Franco Angeli, 2006.
3. Cerruti Mainardi P, Vianello M G, Bonioli E, "Considerazioni su 5 casi di sindrome di Cri du Chat.", Minerva Pediatri 1976; 28:2389-400.
4. Cerruti Mainardi P, "La sindrome del cri du chat in età adulta." In: Andria G, Dagna Bricarelli F, Del Porto G, De Marchi M, Federico A, "Patologia genetica ad esordio tardivo.", Monduzzi, Bologna, 1987:113-28.
5. Cerruti Mainardi P, Pastore G, Guala A, "Sindrome del Cri du Chat." In: "Linee guida assistenziali nel bambino con sindrome malformativa." Balestrazzi P, Milano: CSH: 1994: 75-90 6.
6. Cerruti Mainardi P, Calì A, Guala A, Perfumo C, Liverani M. E, Pastore G, Overhauser J, Zra F, Pierluigi M, Dagna Bricarelli F, "Clinical and molecular characterization of 80 patients with 5p deletion: genotype –phenotype correlation." , J.Med Genet 2001; 38: 151-158.
7. Cerruti Mainardi P, "Cri du Chat syndrome." , Orphanet J Rare Dis. 2006 Sep 5;1:33.
8. Cerruti Mainardi P, Guala A, Pastore G, Pozzo G, Dagna Bricarelli F, Pierluigi M, "Psychomotor development in Cri du Chat Syndrome." , Clin Genet. 2000 Jun; 57(6):459-61.Clin Genet 2000 Aug; 58(2):156.
9. Collins M S, Cornish K, "A survey of the prevalence of stereotypy, self-injury and aggression in children and young adults with Cri du Chat syndrome." , J Intellect Disabil Res. 2002 Feb;46(Pt 2):133-40.
10. Cornish K, Bramble D, "Cri du chat syndrome: genotype-phenotype correlations and recommendations for clinical management." , Dev Med Child Neurol. 2002 Jul;44(7):494-7. Review.
11. Cornish KM, Bramble D, Collins M, "Cri du Chat Syndrome. Guide Lines for Parents and Professionals." , Cri du Chat Support Group, 1998.
12. Cornish KM, Munir F, "Receptive and expressive language skills in children with cri du chat syndrome." , J Commun Disord. 1998 Jan-Feb;31(1):73-80; quiz 80-1.
13. Cornish KM, Pigram J, "Developmental and behavioural characteristics of cri du chat syndrome." , Arch Dis Child. 1996 Nov;75(5):448-50.
14. Estivill E, De Bèjar S, "Fate la nanna." , La Mandragora s.r.l., Firenze, 1999.
15. Evans Morris S, Dunn Klein M, "Pre-Feeding Skills: A Comprehensive Resource for Mealtime Development." , Therapy Skill Builders, 2000.
16. Kondoh T, Shimokawa O, Harada N, Doi T, Yun C, Gohda Y, Kinoshita F, Matsumoto T, Moriuchi H, "Genotype-phenotype correlation of 5p-syndrome: pitfall of diagnosis." , J Hum Genet. 2005;50(1):26-9. Epub 2004 Dec 16.
17. Cerruti Mainardi P, Perfumo C, Calì A, Coucourde G, Pastore G, Cavani S, Zara F, Overhauser J, Pierluigi M, Bricarelli F D, "Clinical and molecular characterisation of 80 patients with 5p deletion: genotype-phenotype correlation." , J Med Genet. 2001 Mar;38(3):151-8.

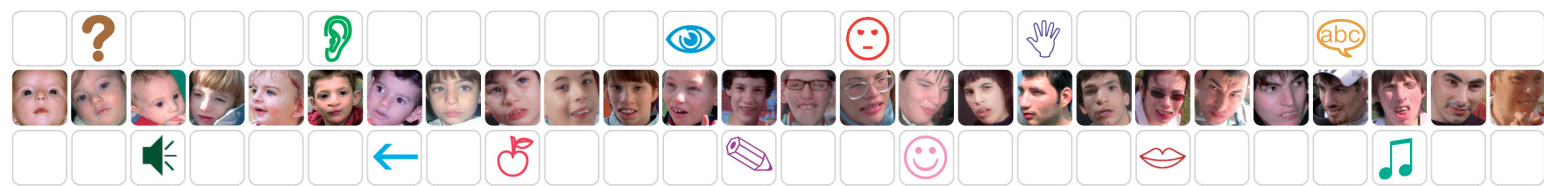


18. Cerruti Mainardi P, Pastore G, Castronovo C, Godi M, Guala A, Tamiazzo S, Provera S, Pierluigi M, Bricarelli F D, "The natural history of Cri du Chat Syndrome. A report from the Italian Register." , Eur J Med Genet. 2006 Sep-Oct; 49(5):363-83. Epub 2006 Jan 13.
19. Niebuhr E, "The cat cry syndrome (5p-) in adolescents and adults." , J Ment defic Res 1971; 15: 277-291.
20. Niebuhr E, "The cri du chat syndrome. Epidemiology, cytogenetics and clinical features." , Hum Genet 1978; 44: 227-275.
21. Pizzamiglio M R, Piccardi L, Massarelli C, Leoni S, Boschetti V, Storaco R, "Protocollo di trattamento neuropsicologico per bambini con sindrome del cri du chat." , I quaderni n.19 I.R.C.C.S. Fondazione Santa Lucia Roma: erre s.r.l. dicembre 2000.
22. Sangalli A L, "L'attività motoria compensativa." , 2006, Trentouno edizioni.
23. Sangalli A L, "L'inserimento dei bambini affetti dalla sindrome del cri du chat nella scuola dell'obbligo. Metodiche operative e strategie didattiche. Educazione sociale e programma intellettuale." , available at: http://educare.it/Handicap/intro/cri_du_chat.htm#udito .
24. Sohner L, Mitchell P, "Phonatory and phonetic characteristics of prelinguistic vocal development in cri du chat syndrome." , J Commun Disord. 1991 Feb;24(1):13-20.
25. Torres C P, Borsatto M C, de Queiroz A M, Lessa F C, Orsi I A, "Cri du Chat syndrome: a case report." , Spec Care Dentist. 2005 Nov-Dec;25(6):286-8.
26. Tullu M S, Muranjan M N, Sharma S V, Sahu D R, Swami S R, Deshmukh C T, Bharucha B A, "Cri-du-chat syndrome: clinical profile and prenatal diagnosis." J Postgrad Med. 1998 Oct- Dec;44(4):101-4.
27. Zhang A, Zheng C, Hou M, Lindvall C, Li K, Erlandsson F, Björkholm M, Gruber A, Blen- now E, Xu D, "Deletion of the Telomerase Reverse Transcriptase Gene and Haploinsufficiency of Telomere Maintenance in Cri du Chat Syndrome." , Am J Hum Genet 2003;72: 940-8.
28. Zhang X, Niebuhr E, Snijders A, Segreaves R, Hamilton G, Brown N, Blackwood S, Hindle K, Niebuhr A, Xiuqing Z, Gray J, Yang H, Albertson D, Bolund L, Pinkel D, "Association of Cri du Chat Phenotype with DNA copy number abnormalities measured by array CGH." , Am J Hum Genet 2005.

This work is dedicated to all those who are dealing with this rare disease, the Cri du Chat Syndrome (SCDC), and need to know more about it.

The Cri du chat syndrome is a very rare genetic disorder that causes a delay in the psychomotor and intellectual development. There is no cure for this condition, but treatments (including speech therapy, physiotherapy and occupational therapy) can help the child to reach their full potential.

This is a very helpful publication that contains plenty of suggestions to help you keep a positive outlook and find the right support if you need it.



This is a non-profit project, if you like it, support it!

www.criduchat.eu



A.B.C. IS A MEMBER OF



A.B.C. HAS COLLABORATED WITH

