THE CRI DU CHAT SYNDROME

A.B.C.
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A.B.C.

The Cri du Chat Children’s Association
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THE CRI DU CHAT CHILDREN’S ASSOCIATION

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Gianlorenzo, my son, my firstborn,
was a real surprise.
When he was born he looked like a porcelain doll,
he was so beautiful.....

but despite everything I’m an optimist
and I like playing and looking at things from a different angle....

so when I cuddle and kiss my son,
I think of him as a four-leafed clover....
because when we search for a lucky charm in a field of clover
what we are looking for
is really a joke dreamed up by nature:
an unusual four-leafed clover instead of a normal three-leafed one.

So our son is a four-leafed clover....
but he can be one only if everybody thinks of him in this way
and not just us.

Gianlorenzo’s mummy
THE CRI DU CHAT CHILDREN’S ASSOCIATION (A.B.C.)

A.B.C. was founded in San Casciano Val di Pesa near Florence in Italy in November 1995, thanks to Maura Masini, the mother of Timothy. She had received little information on her son’s illness when it was diagnosed, and so decided to get in touch with other families who shared her own problems.

The purposes of the Association were to try to eliminate the families’ feeling of isolation by allowing them to talk about their experience and to exchange useful advice; to increase the knowledge of the children’s difficulties so that health-care providers would be better prepared to deal with them; to make public opinion and the medical services aware of the needs of the children’s families; to keep both the families and the care-givers informed of all new developments.

Since November 1995 much progress has been made thanks to the precious work of many voluntary helpers and professional people: at the moment the Association consists of almost 130 families, and the Italian Registry of Cri du Chat Syndrome, set up in the 1980’s by Prof. Paola Cerruti Mainardi, contains data on more than 200 children. Many of the aims of the Association have been achieved; there have been numerous Family Meetings and the study of the Syndrome has been promoted and financed.

A research project has been carried out thanks to the support of A.B.C. on a large number of Cri du Chat children, using the most recent techniques in cytogenetic-molecular analysis (FISH) and the collaboration of both national and international specialists. The project was
also supported by Telethon Italia, and the results have been presented at the most important Genetics Congresses in Italy and the United States, and the Cri du Chat Syndrome Support Group in the United Kingdom.

The studies published up to now regard the evaluation of the psychomotor development in a large group of Italian Cri du Chat children, and have permitted the elaboration of a chart which describes the specific development of the children, information not previously available. An international study has managed to obtain specific development curves for weight, height and skull size.

The results of the study on the genotype-phenotype correlation of a large number of Cri du Chat children have shown a correlation between the clinical severity and the size and type of the deletion, and are important in a practical way because they permit far greater precision than before in diagnosis, treatment, rehabilitation and genetic counselling.

All the information collected from this and other ongoing research is of great help to pediatricians, genetists, health-care providers and especially to the families.

This collaboration is the result of the efforts of the A.B.C. Association to coordinate the work of specialists and families. The bond that has been created and which becomes stronger with time, the gratitude and appreciation of the families, the feeling of hope that is so different from past despair are all comforting and stimulating factors that continue to make the work of the Association worthwhile.
<table>
<thead>
<tr>
<th>INDEX</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>THE CRI DU CHAT SYNDROME</strong></td>
<td>1</td>
</tr>
<tr>
<td>Genetic aspects</td>
<td>1</td>
</tr>
<tr>
<td>Medical problems</td>
<td>3</td>
</tr>
<tr>
<td>How a Cri du Chat child grows up</td>
<td>5</td>
</tr>
<tr>
<td>Psychomotor development</td>
<td>6</td>
</tr>
<tr>
<td><strong>REHABILITATIVE AND EDUCATIONAL THERAPY</strong></td>
<td>7</td>
</tr>
<tr>
<td>Physical therapy</td>
<td>7</td>
</tr>
<tr>
<td>Language and communication</td>
<td>9</td>
</tr>
<tr>
<td>Sleeping difficulties</td>
<td>9</td>
</tr>
<tr>
<td>Behaviour and hyperactivity</td>
<td>10</td>
</tr>
<tr>
<td><strong>OTHER PROBLEMS</strong></td>
<td>12</td>
</tr>
<tr>
<td>Dribbling</td>
<td>12</td>
</tr>
<tr>
<td>Dental problems</td>
<td>12</td>
</tr>
<tr>
<td>Constipation</td>
<td>13</td>
</tr>
<tr>
<td>SOCIAL RELATIONSHIPS</td>
<td>13</td>
</tr>
<tr>
<td>----------------------</td>
<td>----</td>
</tr>
<tr>
<td>Autonomy</td>
<td>13</td>
</tr>
<tr>
<td>Good manners</td>
<td>14</td>
</tr>
<tr>
<td>School</td>
<td>14</td>
</tr>
<tr>
<td>Sport</td>
<td>15</td>
</tr>
<tr>
<td>Parents and siblings</td>
<td>15</td>
</tr>
<tr>
<td>Help for the families in looking after the child</td>
<td>15</td>
</tr>
<tr>
<td>After school what future?</td>
<td>16</td>
</tr>
<tr>
<td>Thoughts</td>
<td>16</td>
</tr>
<tr>
<td>REFERENCES</td>
<td>18</td>
</tr>
<tr>
<td>KARYOTYPE</td>
<td>22</td>
</tr>
<tr>
<td>GROWTH CHARTS</td>
<td>23</td>
</tr>
<tr>
<td>PSYCHOMOTOR DEVELOPMENT CHARTS</td>
<td>33</td>
</tr>
<tr>
<td>TREATMENT GUIDELINES</td>
<td>34</td>
</tr>
</tbody>
</table>
THE CRI DU CHAT SYNDROME

Cri du Chat is the name the French genetist Jerome Léjeune gave in 1963 to a syndrome recognizable from birth due to the characteristic cat-like cry. The word “syndrome” indicates the presence, in the same patient, of different alterations which all stem from the same cause. In this case the cause is the loss (deletion) of a part of the short arm of one of the chromosomes of the fifth pair.

It is a rare genetic condition, with an incidence ranging from 1:15,000 and 1:50,000 live-born, although it is one of the most frequent autosomal deletion syndromes in humans.

The new-born babies tend to have similar facial characteristics: round face, eyes that seem wide apart because of the epicanthal folds (a fold of skin at the inside angle of the eyelid), a small jaw and a retracted chin (microretrognathia). Other features are low birth weight, hypotonia (weak muscle tone) and impaired suction. Over time the face changes: it becomes long and narrow, divergent strabismus is frequent, hands and feet are small. Height and weight are generally below normal, and the voice retains the particular acute tone.

The loss of a part of the 5p chromosome, while producing these effects that taken by themselves would not be remarkable, unfortunately also causes alterations in brain development, with consequent microcephaly (head circumference smaller than normal) and a delay in psychomotor development.

Individuals present a clinical variability in part correlated to the size and type of deletion. In every case early rehabilitative and educative therapies are essential.

Genetic aspects

The DNA is present in every cell nucleus and contains the
genetic inheritance. During cellular multiplication it coils and makes up the chromosomes that are organized in 23 couples, each of which is made up of one chromosome of maternal and one of paternal origin. The couple of chromosomes that determines the sex is XX in the female and XY in the male. The other couples are identified by numbers from 1 to 22. The Cri du Chat syndrome is due to the loss (deletion) of a fragment of the short arm of one of the number 5 chromosomes (5p-) (see page 22). The deletion can be in the terminal part (a single breakpoint) or the inside (interstitial) (two breakpoints) of the short arm. In some cases the deletion derives from other chromosomal rearrangements such as unbalanced translocation, mosaicism, inversion or ring chromosome.

In most cases the deletion results from a “de novo” mutation, as the parents have a normal karyotype. In 10-15% of cases one of the parents is a carrier of a chromosomal abnormality (which may be present in several generations of the same family), more often a translocation, which does not alter his/her genetic inheritance (the rearrangement is known as “balanced”) but can give rise to the deletion in the offspring. These events are not predictable and the syndrome cannot be “blamed” on anyone.

The risk of recurrence is not higher than that of the general population when the parents’ chromosomes are normal. In the case of balanced translocation in one of the parents the risk factor varies from 9 to 19%, as an Italian study has shown: a specific test on the family permits the evaluation of the risk in each individual couple. The prenatal diagnosis is possible and, in any case, genetic counselling provides all opportune information.

Cytogenetic and the more recent molecular-cytogenetic analyses suggest the presence of the “critical regions” which,
if they are included in the deletion, are responsible for the typical aspects of the syndrome.

Two genes, Semaphorine gene (SEMAF) and δ-catenine (CTNND2), which have been isolated and mapped in the critical regions, could be involved in brain development. A recent study in CdCS patients suggests that the deletion of telomerase reverse transcriptase gene (hTERT), which maps in the distal part of 5p (5p15.33), could contribute to the phenotypic changes in CdCS.

The results of studies on genotype-phenotype correlation, for which data was scanty and conflicting, conducted on a large number of Italian children, have confirmed a clinical and cytogenetical variability and the correlation between the gravity of the illness and the type and size of the deletion. These results are of great practical importance because they permit a more precise evaluation of each patient, useful for diagnosis, prognosis and rehabilitation.

Medical problems

In the neonatal period respiratory difficulties (asphyxia, cyanotic crises) are possible, together with impaired suction. These problems can usually be treated and resolved in the pedriatic ward, and transfer to an intensive care unit will only rarely be necessary.

Malformations may sometimes be present: although not frequently observed the most severe are cardiac and renal abnormalities. Less severe are inguinal hernia, syndactyly (fusion of one or more fingers), congenital luxation of the hip, flat foot or club foot, hypospadia, cryptorchidism (testicle in the inguinal canal). Neonatologists and paediatricians will be able to advise on these problems and any further specialist examinations, if necessary.
Life expectation is good and some patients are over 60. The average mortality rate is about 10%, mainly in the first year of life.

Intestinal and respiratory infections are frequent, but only in the first years of life. An Italian research study, evaluating immunology in Cri du Chat children, showed no significant alteration in their immunity defences. It is important that they should have all the recommended and compulsory vaccinations.

There may be some eye problems (divergent strabismus, myopia, cataract, abnormality in the optic nerve) and orthopaedic ones (flat foot, scoliosis) which will require specialist advice. Since there have been some references (infrequent) of patients with neurosensory deafness and the presence of delay in speaking, an audiometric examination is necessary for all Cri du Chat children. Larynx malformation (small, floppy) can sometimes cause anesthesiological problems (intubation difficulties). However, many Italian patients have undergone total anesthesia without complications.

Neurological problems are represented by hypotonia (low muscular tone) in the first years of life, later substituted by hypertonia (improved muscular tone). The microcephaly reported at birth becomes more evident over time. Convulsive crises are rare at all ages. Instrumental examinations (electroencephalogram, CT scan and/or magnetic nuclear resonance) can be useful in single cases.

The seriousness of the clinical situation varies, however, as does late development. Sexual development is generally normal in both sexes.

As with other children, it is the paediatrician who will evaluate the patient’s progress and who will suggest any additional tests.

Treatment guidelines are on page 34.
How a Cri du Chat child grows up

Cri du Chat children at birth are often underweight, shorter and with a smaller head circumference than the norm, indicating that their pre-natal growth was slower. Their weight-gain after birth is also slower than that of other babies.

A baby’s growth is influenced by genetic factors, alimentation necessary to monitor growth in order to prevent any future debilitation that might endanger it. Comparison with healthy children is misleading because children with genetic syndromes have a different growth from the general population. It is therefore more useful to compare height, weight and skull size of Cri du Chat children with specific reference values for this syndrome. For this purpose a multicentre international study has collected anthropometric data of 374 patients: 167 Americans, 150 Italians, 47 British and 10 Australians, which have been used to elaborate specific growth charts for Cri du Chat children (see pages 23-32).

Results have confirmed the existence of pre-natal and post-natal growth retardation: 50% of the children have weight and head circumference values near or below minimum normal values at every age. Height is less affected than weight in the first two years in both sexes, and also later on, especially in males. In adult age, however, the patients are generally short and present microcephaly.

The slowness of weight gain in the first two years may be due to the sucking and feeding difficulties that have been frequently reported. But the fact that this difference between weight and height values continues into the older age groups corroborates the clinical observation that Cri du Chat children are generally slim because of constitutional factors deriving from the syndrome. An awareness of this particular growth
curve in Cri du Chat may help to avoid unnecessary medical tests.

These specific growth charts for the Cri du Chat syndrome, obtained from a large number of children and until now unavailable, together with normal growth comparative charts, will be useful for paediatricians, family doctor, health-care providers and parents to monitor the growth of these children by following documented guidelines.

Psychomotor development

In most cases psychomotor development of Cri du Chat children is very retarded; however this varies between individuals. Few data are available in the literature to evaluate such retardation. In recent years the possibility of studying a large number of Italian children and adults suffering from Cri du Chat syndrome has resulted in specific information on their psychomotor development. The Denver Developmental Screening Test II (DDST II) enables paediatricians to obtain the percentile distribution of patients based on the age of achievement of the various milestones. The percentile calculation allows a rapid comparison of a given patient with both Cri du Chat children and the general population.

The developmental chart thus obtained (see page 33) highlights individual variations, but also confirms that although there is a notable difference from normal children, 50% of the patients can stand with help at 21 months, and can walk by themselves at 3 years of age, and as many as 95% manage to walk before 8 years of age; while 50% are capable of holding objects at 9 months and can feed themselves at 4 years of age. With regard to language development, particularly slow in Cri du Chat children, it has been noted that 50% of children say
“mummy and daddy” at 3 years of age and manage to combine two different words at around 5 years of age.

Surveys have demonstrated that Cri du Chat children, although they have a severe development delay, if home-reared with the opportunity of following rehabilitation from a very early age, can achieve many skills during growth and continue to learn as adults.

The achievement of development milestones should be evaluated in all Cri du Chat children to give a more accurate prognosis to the families and specific guidelines for the health-care providers.

It is useful to keep a diary of the child’s psychomotor development in order to monitor his progress.

**REHABILITATIVE AND EDUCATIONAL THERAPY**

**Physical therapy**

The Cri du Chat syndrome cannot be cured by medicines or operations. The deleted DNA segment can never be recovered, and in any case brain damage occurs in the first weeks of pregnancy. However, it is possible to treat the consequences of the genetic alteration through the rehabilitative therapy which should begin in the very first weeks of life. Its success depends on parents and health-care providers working together.

Several rehabilitation techniques have been evolved and it is important to choose those methods best suited to each individual are useful for Cri du Chat children.

Parents should ask an infantile rehabilitation specialist to teach them the necessary techniques to improve sucking and
swallowing (these feeding difficulties can contribute to delay in growth).

Muscular hypotonia (frequently present) makes the child “floppy” and less active than other children: this leads to a delay in “keeping the head steady”, sitting alone and walking. Coordination of movements can be stimulated by appropriate exercises which help to contrast the pull of gravity thus allowing the child to sit upright and gain autonomy.

The child must then be encouraged to move by himself using every possible means, although the exercises should be carefully adapted to the severity of his symptoms.

When he begins to smile and relate to his surroundings it is necessary to stimulate his interest both visually and orally so as to teach him to pay attention. The stimulation must not disturb him: the best way is to catch his eye and to speak softly.

In order to avoid chewing problems it is advisable to pass gradually from smooth blended food to finely minced and then chopped-up food. It is best to allow the child to eat with his hands from a very early age, as this is an excellent exercise for eye-hand coordination. It is highly motivating, variable and is repeated several times a day. In this way the child manages to accomplish a difficult task without being aware of it.

It is a good idea to start toilet training as early as possible. Put the child on his potty for a short time (some minutes) when he wakes in the morning and after each meal. The parents must not show disappointment but continue to encourage the child patiently, making no comment when nothing happens but praising the child when he is successful.

It is not a good idea to make the child sit still at other times, because the sitting position is not conducive to movements which the child should be learning.

However, contact with an infantile rehabilitation specialist,
as soon after the birth as possible, is of the utmost impor-
tance; as is the collaboration between the family and the
health-care centre. The specifically devised programmes prep-
ared by the centre can be carried out by the family at home,
under the periodic control of the health operators.

Since all Cri du Chat children present clinical variability, as
proved in the most recent research papers, they need to follow
individual rehabilitation programmes.

Language and communication

Cri du Chat children are particularly slow in verbal develop-
ment and require the constant help of their family and ther-
apist.

A recent study has shown speech comprehension to be
notably higher than the ability to communicate.

Logopaedic treatment cannot modify the typical voice char-
acteristics, but can help patients by improving clarity of
expression. Alternative well known language forms can be
used with those children who cannot manage to communicate
verbally.

Sleeping difficulties

Sleeping problems are extremely common in Cri du Chat
children, especially if they are hyperactive. It should be
remembered that all children cry during the night, and it may
be useful to let them cry a little, before rushing to comfort
them, in the hope that the crying will succumb to sleep.
Parents can help the child to sleep well by establishing a rou-
tine that covers the entire day, with meals and bedtime at set
times. It is easier to fall asleep from 8.00 p.m. to 8.30 p.m. in
winter and from 8.30 p.m. to 9.00 p.m. in summer.

The child’s room should be quiet, dark and comfortable. It is his parents’ job to teach him how to fall asleep, by keeping calm and patient and by singing him lullabies or telling fairy tales. Then after saying goodnight they should leave the child alone, even if he cries or tries to attract their attention.

This technique does not produce negative effects, and after a few nights it can help reduce the parents’ stress levels and improve the child’s behaviour during the day.

If however the parents cannot bring themselves to ignore their child’s cries during the night there is an alternative method. They can teach him to be alone by degrees: first by lying beside him while he falls asleep for some nights, then by sitting on his bed, then by standing near the door…. This usually works, but obviously takes more time than the previous method.

It should be remembered that an intensive day’s play in the open air is the best way of making the child feel sleepy.

On the other hand Cri du Chat children are very emotional and they can be troubled by external tensions. During the night they wake easily if they hear a noise because of their highly sensitive hearing. It is a good idea to explain these noises to the child so that he will be able to recognize them and not be frightened by them: we are not afraid of things that we know.

Other suggestions of how to overcome sleep problems can be found in the Bibliography.

**Behaviour and hyperactivity**

Cri du Chat children are often hyperactive, restless, impulsive and reckless.

A research study has shown that hyperactivity is considered the most serious behaviour problem and that it is present main-
ly in patients with a relatively high intellect. Fits of bad temper and self-injury are less frequent problems, found more often in patients with a relatively low intellectual level.

However, not everything is a problem. Cri du Chat children are usually happy, affectionate and sociable. They are often restless and hyperactive when they are stressed or in a bad mood, or when they are trying to attract an adult’s attention.

Hyperactive behaviour can be prevented or controlled by following general rules, such as good sleeping habits or an affectionate but decisive education system.

If the child is behaving dangerously or impossibly and pays no heed to the parents’ instructions (given with decision, but calmly), he must be forced to stop. This educational method must be clear and constant so that the child will be able to understand that such behaviour will not get him what he wants and he will not receive attention until he has calmed down. This naturally infers that when he behaves well he must be praised.

To get round absent-mindedness and distractibility it is important to communicate with the child by direct eye-contact and by using concise and clear sentences together with explanatory gestures. This is essential in order to be listened to and understood. At such a point the child can be given simple tasks to perform, which require a low level of attention and concentration, and which can very gradually be substituted by more complex tasks.

Cri du Chat children can sometimes put on a display of bad temper. These tantrums (such as biting himself, hitting his head with his hands, using his head as a battering-ram) or stereotypes (repetitive movements without any real purpose, like clapping hands or waving them about) can happen when the child is bored or anxious or absorbed in a particular activity. Telling him calmly to stop and giving him something else to do which will
keep him busy is a way of getting round the problem. Since these children are hypersensitive to noise whispering is useful.

Upbringing of these children, who must be taught just like any other child, can improve difficult behaviour. This very hard task must be supported by the advice of paediatricians and neuropsychiatrists. These problems tend to diminish as the child grows up.

Medicinal therapy should be taken into consideration only in particularly serious cases, as results are often unsatisfactory and can cause side effects. In any case such a therapy should be administered only if advised by a child neuropsychiatrist.

OTHER PROBLEMS

Excessive dribbling

Most Cri du Chat children suffer from excessive dribbling which, in many cases, means frequent changes of clothes during the day. This dribbling is due to poor swallowing control and can be improved by logopedia and physiotherapy started at an early age. Special facial exercises can also be useful.

For particularly serious cases an operation to move the salivary glands has been suggested, but work has still to be done on possible future side effects.

Dental problems

Observation carried out on numerous Cri du Chat patients did not reveal a high level of dental decay; and oral hygiene was good in milk teeth. But in older children the conditions were worse, especially with regard to recurring gum disease.
The “open-bite” malocclusion was frequently observed. These results pinpoint the need for continuative dental therapy, usually simple but carried out regularly (sessions of professional dental hygiene). There is no reason why more complex therapies cannot be carried out, as long as a local anesthesia is used. It is necessary to obtain the child’s collaboration in daily dental hygiene, which may not be easy, so this chore should be carried out in the presence of the parents or with their help. In children under 12 fluorine tablets have been proved useful. Hard work, also in this area, can bring about satisfactory results.

**Constipation**

This problem has been frequently referred to by the parents of Cri du Chat children. Potty training, which establishes a routine, is very important. Also important is a diet rich in fibre: fruit (especially prunes), vegetables and wholewheat cereals. It is not a good idea to use laxatives on a regular basis, although sometimes the paediatrician or family doctor may recommend an enema.

**SOCIAL RELATIONSHIPS**

**Autonomy**

It is important that a Cri du Chat child should achieve as much autonomy as possible. It is natural that the family should instinctively want to “protect” him if he is in difficulty, but this behaviour will impede or slow down
his attempts to become self-sufficient. He will be encouraged and gratified by anything he manages to do by himself.

The family should teach him to dress and undress without help, to use at least a spoon and fork correctly, to go to the toilet by himself and to do other tasks; thus reducing his dependence on others as much as possible.

Good manners

The Cri du Chat child lives in a social context, and the people who surround him do not always have to adapt to his wishes. Although he obviously has particular needs he must learn to behave himself when he is with other people, both at home and outside. The rules learnt at home will be automatically applied to the new surroundings and will act as a help towards good manners: say hallo, don’t be a nuisance, don’t interrupt, answer the question...

School

Early attendance at infant school is not recommended as the child has not yet learnt social graces and would be exposed to respiratory infections.

However, nursery school is advisable because contact with other children will teach the rules of social behaviour much more effectively than the family is able to.

The family, teacher and therapists must all work positively together in order to support the child in whatever school he attends. It will be necessary to devise individual education programmes that can be adapted to the child’s learning level.
Sport

Work-outs in the gym, football, swimming, horse-riding, mountaineering or skiing are fun, and also helpful ways to acquire experience. They are moments that the children and their families can enjoy together.

Parents and siblings

The presence of Cri du chat children in a family means a constant commitment on different levels which is far-reaching. Each family member is bound to be affected. A study of families with Cri du Chat children has concluded that the internal stress levels are linked to the degree of behavioural problems of the children. Any help from relatives, friends and specialists should be gratefully accepted. But the reactions of healthy siblings and their ideas about their Cri du Chat brothers and sisters are not those generally supposed by their parents, who consider their healthy children much more preoccupied and unhappy than they actually declare themselves to be.

However, psychological problems can often be present in parents and siblings, and a session with a psychologist can prove helpful.

Help for the families in looking after the child

Sometimes people who are not part of the family home circle may help look after him for short periods of time, varying from a few hours a day to several weeks.

It depends on the area of residence. All over Italy voluntary work is very active and helpers are invaluable and much appreciated by many families.
In some regions the Town Council organizes short holidays for handicapped children in pleasant surroundings.

**After school what future?**

The life of a Cri du Chat child after he has completed compulsory middle school is a constant worry for parents. Before he finishes school it is a good idea for the parents to ask his teachers what they consider could be the best solution for him in the future.

There are various alternatives: training-schools, educational centres, or a job.

The centres (either day or boarding) are run by specialist staff and offer numerous activities that help to develop social and communicative abilities and independence. In the centres’ workshops manual activities are taught, and the centres themselves are willing to collaborate with the family by continuing any prescribed specialist treatment.

**Thoughts**

“Life with a Cri du Chat brother can be: funny, boring, tiring and sometimes quite sad; my life is a bit of all these”.

“Not many people ask me what’s the matter with my brother, but those rare times that they do I’m really happy and I’m very grateful if they let him play with them”.

“Francesco can swim strongly in deep water, and he can ski, run, talk, sing and has a great sense of humour. He is extrovert, gentle and loving, but can also be headstrong and disobedient. All his friends and acquaintances consider
him to be exceptional”.

“Eleonora is great fun to be with, we often find ourselves laughing at something she says or does. She is full of vitality and enjoys being with other people.... she’s also very stubborn and is capable of insisting for days until she gets what she wants. She’s hyperactive and never stops running and jumping around. We love her very much and couldn’t live without her”.

“Riccardo is fifteen: he’s quite slight but strong and tireless, he loves to be in a crowd and his friends are readily available and protective towards him; he likes music, watching television and listening if someone reads aloud to him. The fact that he is so happy, affectionate and out-going makes everything much easier”.

“Angelica is eight and a half and can do a great many things: she can nearly manage to wash herself, she gets dressed haphazardly, she eats with cutlery, helps with the housework and is very tidy. She adores playing with any type of ball, likes going swimming and playing around in the water and is captivated by music”.

“Alberto has some exceptional abilities and a good chance of improvement in the future. He is an affectionate and sensitive boy, very vivacious and curious about what is going on around him. He instinctively realizes that other people are interested in him in many ways, and he is capable of ingratiating himself with everyone.

I’ve been asked what it’s like to be Alberto’s brother, and my answer is that it’s an experience that teaches you life’s true values and that helps you to grow up”.
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KARYOTYPE

Female karyotype showing, at the level of one of the chromosomes of the 5th pair, the loss of the segment of the short arm that causes the Cri du Chat syndrome.
Fig. 1: weight of females with Cri du Chat syndrome from birth to the age of 24 months (blue lines). The normal growth curve (thin red lines) is shaded in orange.

Fig. 2: weight of females with Cri du Chat syndrome from 2 to 18 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.

Fig. 3: weight of males with Cri du Chat syndrome from birth to the age of 24 months (blue lines). The normal growth curve (thin red lines) is shaded in orange.

MALES WITH CRI DU CHAT SYNDROME FROM 2 TO 18 YEARS

Fig. 4: weight of males with Cri du Chat syndrome from 2 to 18 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.

Fig. 5: height of females with Cri du Chat syndrome from birth to the age of 24 months (blue lines). The normal growth curve (thin red lines) is shaded in orange.

**GROWTH CHARTS.6**

**Fig. 6:** height of females with Cri du Chat syndrome from 2 to 18 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.

Fig. 7: height of males with Cri du Chat syndrome from birth to the age of 24 months (blue lines). The normal growth curve (thin red lines) is shaded in orange.

Fig. 8: height of males with Cri du Chat syndrome from 2 to 18 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.

**GROWTH CHARTS.9**

![Chart showing head circumference of females with Cri du Chat syndrome from 0 to 15 years.](chart)

*Fig. 9: head circumference of females with Cri du Chat syndrome from 0 to 15 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.*

Fig. 10: head circumference of males with Cri du Chat syndrome from 0 to 15 years (blue lines). The normal growth curve (thin red lines) is shaded in orange.

PSYCHOMOTOR DEVELOPMENT IN 91 ITALIAN PATIENTS WITH CdCS

Legend

1 = 90% of the normal American population (DDST II), see Frankenburg et al.
• = 90% of the normal Italian population for available skills, see Prina et al.

By kind concession of SIP and Pacini Editore S.P.A.

# Treatment Guidelines

## Problems

<table>
<thead>
<tr>
<th>Neonatal problems</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Low weight at birth, possible respiratory problems (asphyxia, cyanosis)</td>
<td>Neonatal and paediatric treatment, Psychological support to the family</td>
</tr>
<tr>
<td></td>
<td>Difficult suction, vomit, hypotonia</td>
<td>Importance of starting physical therapy (to improve suction and swallowing) right from the first weeks of life, Possible breast feeding</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Congenital malformations (rather uncommon)</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heart (septum defects, arterial duct opening)</td>
<td>ECG, chest X-ray, BD Echocardiography at diagnosis</td>
<td></td>
</tr>
<tr>
<td>Gastrointestinal tract (malrotations, Hirschprung disease)</td>
<td>Abdominal echography whenever necessary</td>
<td></td>
</tr>
<tr>
<td>Brain (agenesis of corpus callosus)</td>
<td>Transfontanellar echography at birth, CT and MNR if indicated</td>
<td></td>
</tr>
<tr>
<td>Kidneys</td>
<td>Urogenital echography at 2 months</td>
<td></td>
</tr>
<tr>
<td>Congenital luxation of the hip</td>
<td>Hip echography at 2 months</td>
<td></td>
</tr>
<tr>
<td>Chryptorchydism, inguinal hernia, syndactyly, cleft lip and palate</td>
<td>Advice from the paediatric surgeon</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Neurological problems</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypotonia followed by hypertonia, psychomotor retardation</td>
<td>Early rehabilitation (right from the first weeks of life): physical therapy, psycomotricity, speech therapy Important a close collaboration between families and operators</td>
<td></td>
</tr>
<tr>
<td>Neurosensory deafness (rare)</td>
<td>Audiometric examination in the first months of life</td>
<td></td>
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<tr>
<td>Convulsive crisis (rare)</td>
<td>EEG</td>
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<table>
<thead>
<tr>
<th>Anesthesiological problems</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Possible difficult intubation due to larynx abnormalities</td>
<td>Anesthesiological consultation</td>
<td></td>
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<table>
<thead>
<tr>
<th>Recurrent infections</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Respiratory and gastrointestinal</td>
<td>Immunological and allergologic evaluation, Compulsory and recommended vaccinations</td>
<td></td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Eye problems</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Divergent strabismus, myopia, cataract</td>
<td>Periodic eye examination</td>
<td></td>
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</table>

<table>
<thead>
<tr>
<th>Orthopedic problems</th>
<th>Clinical data</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flat foot, pes varus, scoliosis</td>
<td>Periodic orthopedic examination</td>
<td></td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Dental and mouth problems</th>
<th>Clinical data</th>
<th>Management</th>
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</thead>
<tbody>
<tr>
<td>Scarce dental caries</td>
<td>Early oral hygiene. Regular sessions of oral hygiene with a professional</td>
<td></td>
</tr>
<tr>
<td>Frequent malocclusion like “open bite“</td>
<td>Possible dental or orthodontic treatments preferably in local anesthesia. Fluoroprophylaxis</td>
<td></td>
</tr>
</tbody>
</table>
THE CRI DU CHAT SYNDROME (edition 2)

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This English translation is dedicated to the memory of Dorrie and Jim Robinson

A.B.C.
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For Timmy
From Benedetta
I HOPE YOU MANAGE
TO UN-HANTICAP YOURSELF!