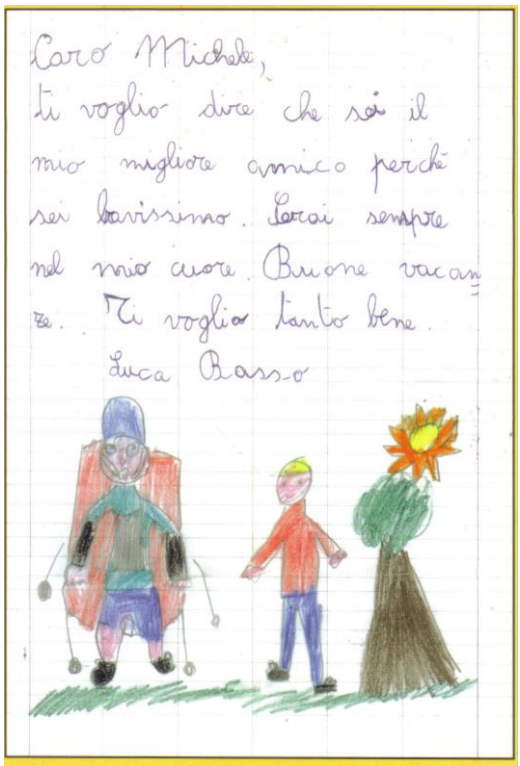
An (Im)Possible Life



Stories of children whose life has been stated "impossible"

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Tales...

It is the end of August at nightfall. The air is clear and tranquil on the beach.
You look at me with your usual, serene, trustful look.
The calm that the rhythmical sound of the waves on the shore and the scent of the sea give are both wonderful, now that the sun has just set.
A seagull is carried on the air, so silent now that most people have gone home.
In your eyes I can see a question. Would you like me to tell you a tale?
You smile.
“Once upon a time there was a little bud, just bloomed like you.
It was morning and everything was bright and cheerful. The little blossom, awake for the first time, was looking around him.
He saw a bee buzzing speedily among the flowers.
He saw and heard a little bird trilling on a branch and.look, it has just flown onto the higher one.
He saw a busy spider building a thin cobweb meticulously between a blade of grass and a wall.
The little blossom was astonished at the movement, the light, the colours.
He was filled with wonder at the sounds.
He felt an uncontainable force, a pushing impetus in his short stem and he thought: “Now I am going to move too, I am going to sing, I am going to do something: I can feel it!”
But when he tried to reach out towards the other flowers on the branch, he discovered that he was not able to move and when he tried to sing the life that was rising inside, no sound came out from his petals.
For a moment he was seized with panic and angst and he thought he was fading.
But right at that moment a gentle breath of wind began to rustle the branch slowly.
The little blossom stirred himself and relished the caress of the wind.

He heard a voice in the passing breeze.
A thin, sweet, beautiful voice.
It was saying: "You are alive. You are beautiful. I love you"
The little blossom was so happy that he forgot his previous angst. He opened up even more and, look, from the deepest of his being an intense scent gushed out of him and a butterfly alighted on him bringing him his neighbours' pollen.".
What a beautiful colour the sky is now, my baby!
There is still light, but it is fair and tender, making our chat at the seaside more intimate.
Perhaps it would be better if I put your shirt on you: do you want to go home?
Let's still taste this peace together for a little longer...
What a wonderful look you have got: shall I tell you another tale, then?
Your marvellous smile on your small, three-year-old child's mouth is an eloquent answer for me.
And then: "Once upon a time, in a beautiful garden, there was a fishpond with gushing water inside and a lot of goldfish..."
Yes, like your goldfish, at home: you laugh at remembering it, don't you, cheeky boy?
"That spring morning a little goldfish opened his eyes and began to swim.
He darted here and there and lifted his head out of the water and saw the thousand drops of tinkling water .
A sunbeam joked with the water and the astonished goldfish saw it changing into a multicoloured cloud.
A dog barked joyfully far away (perhaps his young master had just woken up) and a girl was singing happily for the beautiful day.
The goldfish turned a somersault and almost came out of the water opening his mouth to shout his joy of living.
But "plop" was the only sound he could make.
What? Is that all? But his will to sing, to call out, to talk about his happiness were strong.
Feeling sad, he was about was to plunge into the darkest deep, when the surface of the water rippled in a light tremor because of a breath of wind that began to ruffle the drops of the little water-fall. It seemed to be whispering something.

The goldfish remained on the surface of the water and heard, like in a song, a voice saying:
“You are alive. You are beautiful. I love you!”.
He forgot his discouragement and darted happily towards the other goldfish.”
You like playing with water too, don’t you?
But it is time to wipe your hands and go home!
Another one? I like telling tales, but it is late! All right: it is the last one and then we go home!
“Once upon a time there was a rabbit who got out from his burrow at last.
How beautiful the first walk is: the green grass... a cricket! The flowers. The trees (how high they are!).
The rabbit was jumping here and there, without going too far from his mother. He was curious and delighted about everything that he was discovering; a little afraid of the various rustles, noises and movements filling the meadow. And right in front of him, is a robin.
The rabbit stopped. His heart was beating fast.
The robin turned his little head and he looked at the rabbit in the eyes. He opened his wings and flew away.
How marvellous ! The rabbit immediately tried to follow him.
He tried to swoop into the air but he found himself still in the grass, so he tried again.
The robin sang up there on the branch. The leveret felt sad: he looked at the sun with the leaves high up in the sky joking and longed to have wings to fly up there, too to reach those rays playing hide-and-seek with the robin.
He could not see any higher and turned his eyes back to the ground, to the roots, to the maggots: but everything seemed to be in the shade and his cheerfulness went away.
And again, like in the other stories the Wind came.
He blew through the high branches of the fir-tree, but he also blew down onto the meadow, making the daisies and the grass and the shamrock swing.
He made the rabbit shiver and the cub jumped into the soft coat of his mother.
He cocked his ears because, there, in the Wind he heard a voice speaking to him: ”You are alive. I love you. You are

beautiful!".
The rabbit forgot his sadness and plunged his mischievous little muzzle into the thick warm coat of the hare."
Speaking about warm coats: can't you feel how the air is cooler now?
We should go home! Look, now all the children have left the beach: they have collected their buckets and they have gone away, toddling after their mothers who were carrying heavy bags.
You too were watching them following their fathers who were carrying airbeds and life buoys.
I also saw their little strong legs and their little tanned feet wearing tiny sandals...
Now I pick you up in my arms, my dear child, because you cannot walk. I look into your eyes, because the words that you want to say to me, cannot come out of your mouth yet.
In your intelligent look there is so much vitality and curiosity. In your smile I feel the force of life just discovered.
Carrying you in my arms I walk barefoot on the now cold sand.
A breeze has just started.
Can you feel it?
You close your eyes and lean on my shoulder.
It is strange, I seem to be able to hear a voice in the Wind too: he is speaking to you!
He is telling you: "You are alive. You are beautiful. I love you!".

CHAPTER 1

When an "impossible" child comes into this world: the diagnosis

This short text intends to tell a story and to explain a rare disease, which, despite the prognosis and fears, has not extinguished the wish to live, to fight, to rejoice by many children who are "condemned" by an unlucky diagnosis.

Curiosity

With the wheels of the pushchair sinking into the clear brook water at 1600 Mt. high, under a blue mountainous sky, Michele determinedly throws his long blade of grass into the water: his face expresses a mixture of joy and care. He is "fishing" and he is enjoying himself very much.

Little Giorgio approaches Michele looking at him with interest. Michele smiles at him.

Giorgio looks at me, pointing Michele's little arms in special jeans splints, to avoid bending and asks: "Why?"

"Because in this manner he doesn't hurt himself", I explain.

Giorgio, satisfied of what he is told approaches Michele again and he too tries to catch the imaginary little fish in the brooklet with a blade of grass.

Almost at the end of the morning, Giorgio has made friends with Michele just like his mother has with me. During a pause in the game he looks at me without shame and ventures "How old is he?" "Seven" I answer.

As always the following question, since it was asked by a young boy and not by a grown up, was not "Can't he walk?" but "Why has he still the pacifier?"

"In order not to hurt his mouth" I explain again and Giorgio, satisfied, goes back playing peacefully.

With grown up people it is more and more difficult.

To grown up people it is so hard to understand this so very absurd and unknown illness...

To understand and to explain: I remember the expression of

the pediatrist when, with the diagnosis he was preparing himself to explain to me and my husband what Lesch-Nyhan Syndrome was...

Memories

We were both waiting in front of the doctor: we knew that it was a serious thing, but we had no idea of what it would involve for our gorgeous, delicious child less than 1 year old.
The silence continued, the doctor never raised his eyes, then, at the end, he began to explain, trying to choose the words carefully: a disease which leads to self distruction...children bite themselves, they hurt themselves...there are renal problems...we don't know the causes of this behaviour...it is rare...his life won't be long...
We asked other doctors, friends of ours, to obtain as much detail on the subject as possible. We only received some photocopies in English and not very up-to-date.
It would have been useful to have had some precise addresses: other families with the same experience, centres or doctors who knew something more about the subject, a book or a pamphlet which explained the disease.
We were told that other parents had not even been given a precise diagnosis. For many years they had believed their children to be affected by cerebral palsy and both the parents and the doctors were surprised at the child's incomprehensible behaviours. To some other parents the future of their children was described in a imprecise and incorrect way.

Lesch-Nyhan Syndrome

The Syndrome is caused by a *genetic defect* whereby the human body is not able to produce an enzyme (a substance which carries out a necessary chemical reaction). In this case the missing or defective enzyme is HGPRT and is connected with the purine metabolism.
The lack of this enzyme creates an excessive production of uric acid which causes renal damage, stone formation, gout and which also produces an alteration to the central nervous system function. The possible cause of this alteration has only

recently looked at.
The symptoms: children, still younger than two, present spasticity (they are not able to sit and to walk. They have uncontrolled movements. They are not able to speak well). At different ages, generally between two and five years old, they present a “SELF-DESTRUCTIVE” behaviour.
It is not surprising when physicians let a certain uneasiness show when describing this syndrome: it is very difficult to understand how a conscious person, who is able to “feel pain”, can inflict himself with serious wounds and permanent lesions in a repetitive and apparently uncontrollable way.
From the information learnt by reading the obtained photocopies -with the dictionary at hand- mental retardation and aggressive behaviour have **always** been pointed out as the principle consequences of the disease. Self-injury seemed to go together with the above-mentioned symptoms. In other diseases too and in some forms of autism self aggressive behaviour is in fact present. Up until Michele began to bite the fingers of his little hand, when he was three, knowing the above information, we had hoped that he was “special”. In fact the readiness in trying to learn despite his severe handicaps, the brightness of his eyes, the sweetness and the tenderness that characterise him and make him loveable to most of the people who meet him, did not seem to coincide with what those photocopies reported...
Unfortunately the information we had was not sufficient to teach us like all that the experience of our daily lives has taught us: the compulsive self-injury (not only biting himself as we thought) causes panic and anxiety in children affected by Lesch-Nyhan Disease. But the awareness of their impulse to self-injury and their fear of the pain are not sufficient to stop them. We have felt Michele’s anguish with anguish and we have realised that this dreadful urge is not voluntary: his intelligence and his understanding to have to fight against his own urge make the symptom of the disease even more dramatic.
The information collected later, thanks to some contacts with the U.S. via Internet, confirmed to us that mental retardation is not in itself a characteristic of the syndrome. It also confirmed

that some learning difficulties, connected with the illness, complicate the evaluation of the intelligence of these children who seem more gifted than what traditional descriptions have reported.
The word “aggressiveness” too, does not explain these children properly: the “destructive” urge, as said “not voluntary” and painful for the one who feels it, is sometimes addressed to things or people near the patient. For example: Michele has knocked someone’s glasses off their face or has knocked a glass out of someone’s hand and then has been dreadfully sorry and begged their pardon.

Impossible life?
Apart from the information collected in those first months after the diagnosis, for many years we had not succeeded in finding other information about the disease, nor about possible families in our circumstances.
One day I chanced to read a book concerning genetics and the hopes on gene therapy.
Among the hereditary diseases mentioned, was Lesch-Nyhan Syndrome: finally two lines in Italian!
The sentence was ”...as the terrible Lesch-Nyhan Syndrome, that makes life impossible”: I was astonished and I felt a strong will to rebel against these words.
Michele’s life is *difficult*. His well-being, his joy, his achievements have the price of a constant fight involving him, us and many caregivers, but the word “*impossible*” is not acceptable: hope is necessary for him, for us and for all parents finding themselves up against this diagnosis.
“Resignation” is very different from “accepting” things: the latter does not rule out the fight and it looks at the future. The former is a death sentence for somebody who is still alive!

How many cases do we know?
If we examine the data we now have, the scarcity of Italian patients is surprising when we compare the number of cases found in other European Countries.

CHAPTER 2

“Don’t throw the baby out with the bath water”: the value of a life.

Now, after having listened to other parents with children affected by the same syndrome, after having read the letter sent from Great Britain by Paul’s parents, a child affected by Lesch-Nyhan Disease, I can say that ours is not a peculiar case: the awareness of the great ***value*** of these children’s life is not only the fruit of a choice.
Ian, Paul’s father writes to me: “He is mainly a very happy child and everyone he meets adores him”
Think that he had to have all his milk teeth out, because no other remedy succeeded in avoiding him from biting his lips and he lived in the fear of hurting himself!
When I was a girl I daydreamed about the saying that goes “do not throw the baby out with the bath water”: I imagined the old bathtubs, a rosy chubby child and an old plump inattentive nursemaid... Today I can say that this paradox well suits certain *emotional reactions* to rare diseases, disability and to diverse generic situations in which people can find themselves.
For some the seriousness of this illness, the “diversity” of these children can seem to undo them... Those who exist however, feel, rejoice, fear, fancy, desire, can laugh, joke and love and are a gift for people around them.
“Navigating” through Internet I have recently found two stories of boys affected by Lesch-Nyhan that confirms the richness of positive contributions to the life of others: of Scott, 18 years old, Canadian, a therapist said: “A young one who is an inspiration to those who think that it is too hard to live...That which captures your attention is the fantastic smile of Scott and his unbelievable sense of humour”.
Of Jean-Marc, 5 years old boy, from Manitoba, nicknamed “Smiley” for his bright smile, considered a “charmer”, an operator said: “For his pleasant personality is welcomed wherever he goes”.
Only by being positive, is it possible to face and to manage

this dramatic illness and perhaps, even to conquer it.

The choice

Why have I spoken of “choice” with regards to Michele? Because he did not happen unexpectedly. We did not expect to have a nice, healthy boy who could run and play like all the others. We adopted him knowing fully the diagnosis, feeling a little desperate for him and with him, but because of this, feeling that he was in need of a family who would accept and cure him, more than other boys.

Of course this fact has put us in a privileged situation from the psychological point of view and also because it immediately appeared that Michele was “greater” than his Syndrome...! But as I was saying, the experience of “natural” parents who have been contacted recently confirms the following idea: our children are **not** just “cases” of a rare disease, but, as their friends immediately understand, they are first of all a **person** even though they are ill and, for this reason, they need to be with people who can understand them.

The rehabilitation

This premise may appear to many (I think) as obvious: unfortunately having heard the experiences from other parents does not render it as such. Along with the stories of improvement and support, we have also unfortunately heard stories of refusal, abandonment and above all indifference, discouragement and grief from those who do not know how to face the suffering of their own beautiful children!

If we just look at the child himself, with his age and his needs, we can immediately see that it is not true when doctors say: “We can do nothing”!

There really are ***a lot*** of things we can do, immediately, to recover as much potential and as many abilities as possible.

Michele was lucky to find, in the first months of his life, a rehabilitation Centre that has put into action all the strategies for a possible recovery. This helped him to go to a normal nursery school and then to elementary school and has helped us not to feel **alone.** The Centre has helped us to manage and cope in the moments of panic that we have had following

those first experiences of self-injuring (he was three years old).
The rehabilitation has not only concentrated on the recovery of his motor capacity: (in fact the athetoid, dystonic spasticity that afflicts Michele does not even allow him to sit without adequate supports), but also on the recovery of his capacity to **communicate** which is hindered by the difficulty to formulate words well.
I remember the emotion I felt the first time that Michele put his finger onto the symbols on his “table”: he was still only expressing himself in monosyllables and it seemed that he could understand and wished to say more than we were able to decipher.
The Doctor of the rehabilitation Centre had proposed to try a method of “Augmentative communication”: the method “Bliss”, based on symbols of easy memorising. After several meetings our boy began to try and express not only his discomfort and his more urgent necessities, but also his feelings, his emotions, his thoughts.
How many pleasant surprises we had in those meetings: discovering that even if he seemed to be -because of his disease - a very small boy, Michele was growing. His curious mind was opening up to the world and we could help him to express his growth!
The table now has been put aside because our young boy has learnt to say many words and is learning to write: but it has been a fundamental spring-board for him and us; a bridge on which to meet, suspended between his and our way of communication.

Communicate
It was important for us that he could communicate, but it was also important for us to communicate with people who have had the same experiences. Talking with the doctors and physiotherapists we found ourselves expressing, more than once, this necessity: we wanted to look at the future but did not know what to look for. The information about the disease, like at the diagnosis, was scarce enough, but information about rehabilitation was practically non-existent! What was

best thing to do? The question became dramatic when, at three years old, Michele began to put his fingers in his mouth and to bite them.
In those moments we experienced with him, the **terror**. In the absence of experience and confrontation, we passed rapidly, after the first small injuries, from trying not to care to making "splints" for his elbows. At the beginning we fixed carton rolls of toilet paper to the sleeves of his clothes with sticky tape...
I remember our initial embarrassment and discomfort in having to take him to nursery school dressed in that way: he began nursery school at the same time he began to bite himself. I was careful to **cover** those strange and disquieting things with his smock !
Today, thanks to the documentation we have collected, I know that physical "containment" is the best solution for him and for the others: to be "restrained" in his movements, if the alternative is hurting himself, should not be regarded as a *constriction* for the children having this disease, but on the contrary, as "freedom", because, feeling "stopped" from hurting himself, the child with the Syndrome of Lesch-Nyhan will relax and can participate with more serenity and tranquillity in everyday activities.
For the child, however, who is already anxious, it is **very important** to feel around him the calm acceptance of his instruments of defence: be they gloves (as I have seen in same cases), links or other "instruments"...I have come to know that in the U.S. some caregivers have found, a solution to save the fingers of their patients, protect them with curlers!
Even if to people they seem strange or sometimes inopportune, the "tricks" that function and that cheer up our children **must be proposed and defended**.
I am writing this, because I hope that my experience will serve other families who are facing the same problem: it is not right for each family to learn on their own when there are others of us who can help them!
We who have no difficulty in talking and writing - COME ON! LET'S COMMUNICATE!

CHAPTER 3

The everyday "struggle": Self-injury

Today, I am sitting in front of Filippo, another young boy affected by Lesch-Nyhan disease. I have come here with Michele having made quite a long journey. No two children with the disease live in the same town, unless they are brothers.
The joy and the comfort of being able to meet other parents with the same problems, to compare our children's work at school, to talk about their achievements are not the only emotions of today.
Filippo, with anguished look, is warning his mother that one of his restraints is slipping off his hand.
And there! He is already putting his hand in his mouth. But now his mother has caught his wrist and he can calm down again.
Watching this, I feel all the same emotions I feel every day: a strong desire to rebel against this tremendous "urge" to injure and a great solidarity towards these heroic children who keep their joy of living, their smiles and their good humour, despite the everyday struggle they have to face with their bodies, that rebel against their own will and logic.
I can see my own anguish reflected in Michele's eyes: his smile has disappeared and he is looking at his new friend seriously and with thoughtful expression.
This experience today will give me the opportunity to talk to him about his illness. I will be able to let him know that I understand his urge and that I ***accept*** this absurd situation. I will tell him that we can do it; that we are ready to help him in his every day battles.

An "impossible" loneliness

When I compare my experience to that of other families, I am convinced that the only *impossible* thing about living with a child who has Lesch-Nyhan disease, is *loneliness.*

The comfort of exchanging ideas and talking about this upsetting behaviour, are absolutely ***necessary*** to preserve at least some kind of tranquillity and equilibrium in life. Te last thing we want to do is further burden our ill children with our own feelings of pain and anguish.

It is a real relief to us, the adults, to talk about their inexplicable urges. In this way, problems do not become overwhelming and invincible and this tyrannical symptom of the disease can be won. And for exactly this reason, I believe that the only way the children can get out of their loneliness is by talking to us; letting us know what they are like, *beyond* their suffering and disease; how they feel about their daily fight.

That is not easy to achieve. An American caregiver has said that even those who daily look after these children on a daily basis seem to get enveloped in this feeling of loneliness, too.

It is with this conviction, that through **communicating** with us our children's quality of life will improve, that all of us involved must have a great capacity for listening. We must be attentive.

My child's state of agitation was often due to the presence of *something dangerous close to him*: something with which he could prick or scratch himself or a wall or a piece of furniture he could hit his head, his foot or his arm against.

His state of agitation often **pointed out** the fact that someone was holding him unsafely, or that he was losing one of his protective restraints.

I have seen people scolding Michele because they do not understand him, and then I have been witness to his helpless acts of self-injury. However I also seen his almost triumphant joy when we understand what the problem is and once the source of danger has been removed, he can relax and calm down.

Even for those parents who have laboriously and with suffering, learnt how to live with this strange behaviour and have come to know how to prevent it, it is always very difficult to let people from the outside understand their situation. I think that this is the reason why children affected by Lesch-Nyhan disease are said to suffer a lot when they change their

surroundings and to feel great anxiety in new and different situations.
As far as our experience with Michele is concerned, we can say that if he is at his ease and is surrounded by people who know what his problems are and who can foresee a probable source of danger when they are with him, then we can change his surroundings, we can travel and give him different experiences. Michele has always loved this, and what is more, he is curious and stimulated by new things. He has quieter, calmer moments during his travels than in his daily routine.

The possible "weapons"

I think **listening** is the most important thing if you really want to take care of a person affected by Lesch-Nyhan disease.
Obviously, parents cannot work alone. They need support from the Health Service, social workers, teachers and also from all the people around them, most of whom, unfortunately, still know nothing about the disease.
Of course **dialogue** is another fundamental element, that is necessary to manage this "impossible" disease.
Creativeness is the third winning weapon. As I previously said, parents' "inventions" are varied and all of them are suited to these children's clever imaginations. As they grow up, they become more and more attentive and, and as a consequence, can "see" new ways of hurting themselves. If helped, however, they can also learn to avoid them!
How many different types of gloves, hats, bandages, harnesses, restraints for lips or teeth I have heard of!
Sometimes discussion with other parents is useful to share and copy new ideas, but it is also useful to discover that the fight can be more effective in certain situations, that we can never "lower our guard", that, if our children feel we are fighting along side them in their everyday battles, it will be easier for them than if we make them feel guilty or humiliated. This only adds more pain to their suffering.
Those who have heard Michele cry after biting his lips or hitting his head, know how much anger and frustration mixed with real pain, are in those tears.

An article entitled “Self-Injury in Lesch-Nyhan Disease” which was published in 1994 in the “Journal of Autism and Development Disorders” by Dr. L.T. Anderson and Dr. M. Ernst, was very useful, as an instrument for a “possible life”.
The research refers to an inquiry carried out on 40 American patients, aged 2-30.
What is illustrated in this study is how much this impulse to self-injury depends on different stressful situations of life (even in case of “positive” stressful situations).
According to these observations, when the children are quiet and comfortable they can control themselves better.
Strong emotions, agitation and stressful situations, on the contrary, often cause the outbreak of a crisis.
When I read this article, I remembered Michele’s birthday party at Nursery school.
There was a big cake with candles, all his friends around him were happy and excited. The teachers were careful and attentive and there were several gifts for Michele who was radiant with joy.
His eyes were shining but as he blew out the candles, he burst into tears because he had bit his lip.
He did not hurt himself much, but it was clear that he was very upset for having “spoilt” the party for the others and himself .
Fortunately, the teachers and the devoted school caretaker distracted him and cheered him up with a “It was nothing at all”, and “It doesn’t matter”, and we went on celebrating.
I fully understood the meaning of what had happened after reading the study of Dr. Anderson: I realised that **Calm, Quiet and Patience** are all essential winning weapons.
It is easy to guess what further studies have confirmed: reproaches, punishments and “disapproval” not only do no good, but rather worsen the children’s behaviour!
Encouragement, comfort, solidarity and comprehension on the other hand, really help them to “restrain” the coming-on of a crisis.
I do insist on this fact because my own experience, though limited, has made me believe that the “aggressiveness” the children suffer is caused by incorrect, negative attitudes

towards the disease. In fact, in medical literature, “aggression” is expressed as behaviour directed towards others and as an intolerance of society. I believe that if we forget that Lesch-Nyhan Disease has its own peculiar characteristics, that is, if we consider and treat in the same way as we do with mental retardation, autism or similar mental illnesses, then we are sure to create a vicious circle, which will be very difficult to break. I also believe that although reproaches seem, at first, to help restrain their children from negative behaviour, the psychological stress caused by the feeling of “being guilty” leads children to much more uncontrollable reactions. Despite my views, however, studies and research into the best attitude to use when confronting a child with Lesch-Nyhan Disease, should be carried out with great seriousness and should follow the criteria of the scientific method.

At this point, an observation presents itself: what we have said can improve their life, but it does not solve the problem at all.

If this uncontrollable urge to self-injury is caused by a *chemical* disorder, then what are the chemical solutions?

That is to say: what are the THERAPIES?

“At present, there are no therapies which can solve the problem of their behaviour”, so everyone tells us. **This is not enough for us**. We want to know the results of any research that has so far been made on therapies already used to treat this disease.

A very short time ago we did indeed wonder if there has been any research at all that can give us a valid offensive weapon against Lesch-Nyhan.

Michele, Simone, Luca, Diego, Giorgio, Jeremy, Paul... and all the children we have met are now entrenched at the front and resist heroically. But *when will the reinforcements arrive*?

At last, for the first time, these questions are no longer without hope of an answer.

CHAPTER 4

Living with other people: integration

The Judge responsible for child adoption asks us if we have followed his suggestion of speaking to our doctor about Lesch-Nyhan and, looking into our eyes, asks again whether, now, we would prefer “therapeutical custody” to adoption.
My husband and I feel uneasy and we try to understand correctly. Are there new obstacles - legal or otherwise - that may prevent Michele from joining our family as our son?
The Judge wants to present the case as clearly and correctly as possible: he tells us what he has been told: he warns us that the child “could not have a social life”.
What a tremendous sentence that comes so readily from such a lack of information about this disease!
We felt overwhelmed with the seriousness of the situation: how could we be so self-conceited and stupid to challenge all these judgements of “impossibility”?
On the other hand could that delicious child - influenced by these awful prophecies - be left **alone** to his destiny?
I know that like us other parents have suffered these cruel moments, too. Some doctors proposed to a mother to leave her child in a hospital and to take no further interest in him. Others predicted life as a “vegetable”...

Children’s point of view

A splendid answer to these terrible prophecies is the letter Michele received from a classmate at the end of the first class at elementary school: “You are my best friend because you are very good. I love you”.
In his accurate and multicoloured drawings I recognise all Michele’s “restraints”: his arms in splints, the dummy fixed to his little hat, the belt holding his shoulders to his waist...
Another girl, aged 13, daughter of friends, after having spent a week in the mountains with us, decide to write a long and beautiful letter to Michele in which she says among other things:

“ I am thinking about all people who are not *lucky enough to know you*... I want you to have the possibility of appreciating life even despite your disability, despite your restraints which do not allow you to move your arms, but which permit you to embrace me...And I feel deep admiration for you, because I have everything and I am happy, but *you are happy* even if you are lacking in many things... A lot of people think you are different and I cannot explain why, perhaps because they do not know you and they cannot appreciate you as you are...When I speak to you, you show me attention and interest in what I am saying...Under the peak of your hat two limpid and confident eyes look at me: “Do you think I am sad?” - No, I do not think so, Michele, but how difficult must be life for you always sitting on a wheelchair!...I love you, dear Michele. My friend, I love you!”

Now we know that Michele’s social life is not only possible but also very special; full of deep relationships with children of his age or older: we know he has gained not only our hearts but that of many children, teenagers and grown up people.

He is a friendly, often cheerful and funny companion.

I have found, via Internet, many other children with the same syndrome and have learnt that they too have fruitful relationships. This reports from the people living near them jar with that short, curt sentence, written in the so called medical “manuals”, and that describes the behaviour of children suffering from Lesch-Nyhan - “aggressive behaviour”-...

What is notable is the scarcity of complete information: ***Michele ran the risk of not having a family*** because it was difficult to make a comparison between the information about his disease and the reality.

The possibility of a real social life, taking into consideration the peculiarity of the symptoms, is strongly linked to the “culture” of the community where the child lives. In a community where difference, disability and disease are regarded as things to be exorcised, then here, for a child suffering from Lesch-Nyhan, it will be difficult to “live with others”. But, where there is place for everyone, even if they are different or difficult, where, along with the problems, you pay attention to the **person**, where the value and the richness

of their lives is recognised, these children will have a "possible" life.
This subject could be lengthened further for it poses questions and looks at considerations which go far beyond Lesch-Nyhan's problems. It deals with the very way we consider Life.
It is not for me to examine this topic closely, but I have to say that a lot of people (and I mean the neighbours, the local shopkeepers, the school, the parish, our friends) have given Michele a hearty reception and this fact has, in turn, given me **optimism.** It has induced me to think that the "diagnosis" that talks about an irretrievable "sick society" has to be revised! There is a lot to feel positive about, even though, as when Michele falls into a crisis, it is easier to be frightened by the negative signals than to concentrate on what is constructive...
In Italy, in those places where the insertion of the children into the regular and public school system works well, there is a great opportunity for our children to "test" out the "possibility" of being with other children, and also of being in difficult, stressful situations. Other children can enjoy the friendship, the sense of humour and the joy that our children have to offer along with gaining a knowledge of the limitations and difficulties that exist. And yet, to realise a deep and true integration into society, *cooperation* between parents, schools and careworkers is absolutely necessary.
In fact, it is very difficult to find the right balance between education, based also on "no", and the effort of preventing stressful, emotional situations, which are the sources of self-injury.
Speaking with other parents, we often try to understand what is the best approach for us to take when dealing with "wrong behaviours". How do we make them understand what "correct behaviour" is and make them more responsible, without punishing them for behaviour brought on by the disease?
Comparing our different experiences is the only possible way to comprehend what we can rightly ask of them, what goals we can expect them to achieve or not, for their growth and greater autonomy.

It is obvious that neither we nor the teachers and trainers must be left to face this difficult, delicate task alone. **Support** for the parents, for classes and for other people with whom these children live, is a real necessity to realise true social integration.
However thanks to the experience of children already “grown up” I became acquainted with the fact that many families have fought this terrible daily struggle **alone** in order to conquer the “possibility” of life and achieve serenity for their children.
Sometimes it has meant woe, doubt, disappointment and incomprehension. Certainly, the same thing can be said for every family who has experienced a rare diseases, but this I understood only later...

CHAPTER 5

The intelligence to understand and manage the intelligence....

Having met children with Lesch-Nyhan syndrome in the flesh, I could not help finding the definition “mental retardation” (which often recurs in the description of this illness) as “strange”.
In fact, their vivacious, intelligent faces, their participation in the everyday life with interest and curiosity, their prompt, timely and sometimes “witty” remarks are features which do not go together with the idea of “mental retardation”.
You must take into account that a dystonic child does not have so many chances nor instruments with which to explore real life as a normal child. Moreover, Lesch-Nyhan children are often intentionally “prevented” from moving. They are sometimes subject to strong emotions and above all they are trying desperately to fight against a terribly fierce illness!
Not only are our children’s learning abilities, collected in the few documents available, all very different, but there are diverse learning programmes as well. Some of children have attended Nursery school for a lot more years than normal children. Others, have attended Primary school at the right age. Some of them have started to learn how to use a computer immediately, while others have never been introduced to one.
In Italy there are cases of children with Lesch-Nyhan syndrome who have been put into public schools, while, abroad, they are usually confined to special schools for the disabled.
It is clear that it would be very useful to uncover the real abilities and possibilities of these children, since to underestimate them could *prevent them from having worthwhile experiences,* so essential for improving their chance to communicate and, so, perhaps, to manage their illness. On the other hand, however, to overestimate them could mean *to submit them to unnecessary stress and*

suffering !
That is why it is necessary to establish a reference point in order to ease and encourage the exchange of knowledge and experiences between educators and teachers. However, I cannot always be optimistic and I feel a strong sense of insecurity whenever I think of all the difficulties to be faced, even the less serious pathologies, such as the integration of the disabled into normal schools and the constant delays in the employment of support teachers who are often not specialised and not very motivated.

A research from the U.S.
Also with regard to the subject of "learning", we have found reasonable explanations and relevant answers from an article by Dr. Anderson (the referent of the International Register of the Faculty of Medicine, New York University) which came out in 1992 in the "Journal of Autism and Developmental Disorders, vol. 22, N° 2", concerning research carried out on 42 patients. The article is entitled "Cognitive Abilities of Patients with Lesch-Nyhan Disease".
From the research it emerges how, in the last few years, the "symptom" of mental retardation, as one feature of the illness, has been put in doubt.
In the past, mental deficiency was often associated with illnesses whose symptoms were serious difficulty of movements or self-injuring behaviour; in fact, the standard methods to measure the intelligence quotient and the mental capacities of patients are based on tests which are impossible for subjects with dystonic movements to do. (Think of the games in which you have to fit geometrical figures into the suitable spaces: Michele has always been able to show me the right place with his little finger, but he has never been capable of inserting one of them by himself!).
Moreover, in many cases, children with Lesch-Nyhan syndrome whose symptoms and behaviour were mostly studied by researchers in the past, had not the chance of attending schools: therefore, for a long time, the diagnosis of mental retardation was accepted without hesitation.

However, it is not necessary to go back many years, also in Italy, to find stories of disabled children kept indoors, far away from any opportunity of increasing their experiences, simply because it was absolutely normal to behave in that manner. Consequently, if they were unable to read and write, it was their own fault, since they were lacking in intelligence and ability. It did not depend on their difficulty in finding suitable instruments of communication.

As for the integration of the dystonic disabled, many advances have been made so far. New techniques of communication and up-to-date technological instruments have opened the doors to a re-examination of the deeply rooted assumptions about those who have difficulty in putting something into words or in holding a pen.

On the basis of this recent method of evaluation, the research carried out by Dr. Anderson came to the conclusion that the children with Lesch-Nyhan syndrome cannot be considered as mentally retarded. In other words, the subjects examined have demonstrated to be able to orient themselves in Time and in Space very well, to feel appropriate emotions and sentiments according to a situation, to have a very good memory, to be capable of understanding the plots of novels, films or the rules of various sports, to be able to concentrate.

A number of these disabled children had also adopted strategies to communicate their needs **very well**, although they were unable to make themselves understood with words. Most of them had revealed a strong desire and a great ability to take part in a conversation with friends and grown-ups and therefore they can undoubtedly be defined as **very friendly and pleasant people.**

However, their difficulty in reading and writing at the same level as children affected by similar dystonic disorders is still widespread. It perhaps happens, as I said before, because a specific strategy of learning has never been worked out, nor compared: these children, in fact, are not only limited by their difficulty of movements, but also by their sudden impulse to injure themselves which they are unable to control and that makes them feel a strong anxiety.

School learning often involves a certain amount of anxiety.

Who after all cannot remember feeling anxious at least once at school?

New ways to follow

It is easy to realise how much these children learn from playing, watching TV or enjoying the experiences of everyday life. They do seem, however, to meet difficulties at school, mainly when they are taught by people who are too strict, demanding and not very "reassuring". There are teachers, in fact, who tend to scold them whenever they show some of the peculiar symptoms of their illness.

As a result of this, the possibilities of learning maybe directly connected with the ability to understand and manage the "Syndrome" itself. The cleverer people near to them are, the more opportunities there will be to develop the intelligence of these brave children!

Reading through this book very carefully you cannot help observing how many obstacles must be overcome in order to work out the best learning programmes for our disabled children. You will see that it is very easy to lose heart, not only for parents but also teachers, if we are left on our own. The difference between our children bright eyes (so called by Dr Nyhan) and not so brilliant school results can discourage us and can lead us to think of "mental retardation".

I speak with full knowledge of the facts: some parents, without support and left alone, not believing in the real capabilities of their child, have been unable to develop the child's potentialities and therefore they waste them.....

And not only in Michele's case, but also in many other cases, a lot of parents affirm their surprise at their child's witty remarks and their "impossible" sense of humour. This gives us yet another reason to keep on fighting and to not throw in the towel.

CHAPTER 6

An illness, a lot of symptoms

Comparing my own personal experience with those of other parents that I have met in Italy, I have discovered that one of the common problems is that it is difficult to identify the branch of medicine which is really concerned with this kind of disorder and to which we can make reference in order to help our disabled children to face the innumerable problems linked with their illness.
Moreover, the number of "specialists" consulted have suggested so many different lines of intervention that we are a little uneasy.
Considering that in Italy there are no specialised centres, some children, since their earliest childhood, have been consulted , by paediatrics skilled in metabolic illnesses, others by neurologists, others by nephrologists, others by rheumatologists.....
Others, however, are taken to physiotherapists, speech therapists, psychomotortherapists, or are given a lot of drugs and tranquillisers or are put on rigorous diets, while others have not.
Some of them have psychotherapy and others have had the aid of able dentists who have designed instruments that can limit injuries to their lips and the insides of their mouths.
All of them use "allopurinol" and this is the **only** treatment on which both the doctors and the medical centres seem to agree! (Allopurinol is a medicine which slows down uric acid production, bringing it back to standard levels even in Lesch-Nyhan patients).
I do not have a precise idea how doctors from different specialised fields communicate with one another. I do know, however that, within their own fields, technical meetings, conferences are organised and specialised journals are published in order to communicate opinions. In the case of rare syndromes that involve so many different aspects of medicine, I wonder if it is really possible for them to meet, to

compare and to exchange their experiences and viewpoints. If this is not the case, what can we do, as parents and citizens, to make the knowledge that there is accessible to each of the different medical fields?

Measures of this kind are also essential in order to avoid complications and provide therapies that may be useful in some aspects of health. Consequently, the idea that a specific therapy might not exist cannot prevent us from acting. In fact, any success, however partial it may be, is extremely important since it contributes to the improvement in the quality of their lives. It surpasses the assumption that “nothing can be done”, which is too often associated with the diagnosis.

Our children manifest different problems, sometimes at different times, but “similar” stories, if closely examined, may help to increase the understanding of the syndrome itself.

For instance, the stories told by several parents reveal a common element: their child’s difficulty in breathing connected with collapse. What is the cause of this? Why do some children not show these symptoms? Are such physical reactions strictly linked to the Syndrome or to the drugs that are sometimes administrated to them? Or perhaps a correct “position” taken in the earliest years of life have prevented them from injuring themselves? Some of the children I know have not been immediately provided with adequate highchairs or pushchairs which not only help to avoid dystonic symptoms, but also help to avert self-injuring crises. Almost all the people close to these disabled children keep talking about the “great fear” produced by their children’s uncontrollable impulse of injure themselves.

How many times, for example, have certain “aids” become, unconsciously, a source of great anxiety! (Corners of tables, bolts sticking out of wheelchairs, hard or rough surfaces which are insignificant for most dystonic children, but are very dangerous for those who look at them as potential instruments with which they can injure themselves).

Moreover, some children have been submitted to particular examinations, while others have not been tested at all. Why?

The renal problem is not always pointed out as one of the primary symptoms of this disease, at least not in all cases.

Certain precautions can prevent nephrological complications, but if a child is referred to a neurologist, for example, the child concerned may not be properly informed about this particular precaution...

I believe that no one will be shocked to find his family doctor unprepared when faced with this illness, thankfully so rare. But, I think that everyone would agree with me if I said that our doctors must refer us elsewhere, or let us consult other experts in order to help bring to light all the symptoms which are strictly connected with Lesch-Nyhan syndrome and to take the appropriate measures.

This is the only way to go; encouraging the exchange of experiences and knowledge, trying to create a “competence” to which we can refer, so as to avoid parents from meeting embarrassed doctors who, although in good faith, give a message of “defeat” just when the real battle should begin.

CHAPTER 7

The hope in a therapy

Unfortunately the fragmentary knowledge, the poor exchange of information, the absence of specific reference points do not only have an effect at the present, people who every day live with the uneasiness of hearing “nothing can be done”, but they also impede the construction of a THERAPEUTICAL PROJECT.

We know that there are more than 200 people diagnosed affected with the disease in Europe (and who knows how many people are not diagnosed). We know that the syndrome is caused by the lack of an enzyme (which has been identified and can be produced in a laboratory). The gene causing the disease has been studied for years and physicians have already tried, on laboratory rats, to “adjust” the cells. Some drugs have been tested and they have had very significant effects even if for short periods (for example L-5-Hydroxytryptophan, as referred to in 1979). For these reasons we are astonished to ascertain that **there seems to be no therapeutical prospects** .

A few years ago, when for me, the possibility of having contacts through Association was unthinkable, I believed I could “trust” in medical and pharmaceutical progress, because I thought it was normal in a world where technology , science and medicine take continual steps forward.

Now unfortunately I have to admit that my thought was naive: only that which **leaves**, will **arrive** with time: a real effort in research for a therapy.

The information collected, also via INTERNET has proven to be a very powerful means by which to receive information otherwise reserved for scientists, but has lead me to think that the various results are not the fruit of co-ordinated and multidisciplinary work, but parts of studies on a few patients at a time, and for this reason, subject to various interpretations. The risk is that when a scientist publishes a result, it can be left unused.

If it is so (and I hope to be clamorously denied), it will be very discouraging!

Gene therapy

Recently there has been talked about a new hope of gene therapy for diseases like Lesch-Nyhan. But what does it mean?

This new approach to treat diseases, nowadays successfully applied to some serious pathologies, such as SCID (a rare and dangerous immune-deficiency caused by the lack of ADA enzyme), gets to the root of the illness: for example it corrects the defect of the gene responsible for the production of the enzyme.

For many diseases the first step is to recognise one or more genes responsible and identify them in chromosomes: today only a few thousand of the approximately 50 thousand genes, that make up human DNA, have been identified.

As I already mentioned in the case of Lesch-Nyhan the gene not only has been identified, cloned and sequenced, but there have also been successful experiments in the transfer of genes in laboratory animals.

There still are, however, "technical" problems that, it is reasonable to suppose, can be overcome with research aimed in the right direction. Laboratory work has begun on other diseases linked to the central nervous system that may lead to valid results.

Pharmacological therapy

In June 1996 some interesting articles were published that illustrated the opening of a new technique in research of the nervous system, the PET (Positronic Emission Tomography). A study was made on 10 normal patients and on 6 with the Syndrome of Lesch-Nyhan. Te findings were the same as shown in other studies: **in the brains of the patients with Lesch-Nyhan disease there is present a notable reduction in levels of dopamine.**

This leads us to think that there could be attempted interventions with drugs already on the market: the trials made up to now have not had the possibility to compare

their efficacy like the new research techniques have had.
Above all, with such a low number of cases the only observation of the clinical effects of a drug does not leave much margin for objective evaluations.
Naturally, whether for new pharmaceutical approaches, or for a project for gene therapy, a push for the research in this direction it would be required, because as I said earlier “only that which leaves, will arrive with time”!
It seems to be a vicious circle: few therapeutic prospects produce few cases brought to light and few connections.
The few cases drawn attention to and diagnosed (in Italy there are definitely more than 14 that we know about!) reduce the possibility to organise a push for research.
This circle should be broken, but only the families of the patients ***really*** have the will and therefore it is from them that the battle should start.
There is an other vicious circle: “few patients = few possibilities of finance” can be broken down by those who insist that it is **worth the wile** to “invest” in the resources of everybody in order to improve the quality of life for **everybody**.
The results of the research on all these rare diseases brings benefits also for the knowledge and the cure of the most common diseases.

CHAPTER 8

And moreover, we now come to dialysis

Notwithstanding the difficulties caused by Lesch-Nyhan Syndrome, the life of Michele has been rather serene. We have also seen with satisfaction, the conclusion of his first year of school.
Unfortunately for him the problems have not finished yet.
His analysis began to worsen and one day both my husband and myself found ourselves yet again in front of a doctor who gave us bad news.
The atmosphere brought back the same situation of a few years before: we had in front of us a person in whom we trusted who was telling us that further troubles lay ahead.
Michele's kidneys were going to rack and ruin and soon his conservative therapy, his diet, everything else would become useless.
Moreover, it was improbable that Michele could be submitted to hemodialysis because of all his motor difficulties.
Feeling very upset, we asked ourselves if any possibilities still remained: how precious Michele's life appeared to me in that moment, how "impossible" life would be without him!
The doctor explains: there is a possibility for Michele to undergo peritoneal dialysis. A simple operation to apply a catheter to the abdomen with daily dialysis thereafter.
He then confronted us with the unknown: how would he react to these difficult situations? How would it work? What is the right thing to do for him?
At home the question was put to all the family and was resolved with the decision to face this fight also alongside him with the deep hope to succeed again and to taste every possible moment of his life, up to the end...
Michele has therefore also lived "stressful" situations in the Hospital (for 4 long weeks):he had to submit himself to long hours of immobility, while I learned to assist him in the manual dialysis. I tied up his hands every evening when he was put on dialysis for the night, thanks to an automatic

machine that controls the peritoneal exchanges. In fact we were terribly afraid that he could hurt himself by pulling his catheter.
During all this time he was marvellous. He had his “seedy” days, but, altogether he was very collaborative, patient and encouraging.
He has never shown signs of wanting to hurt himself, but on the contrary, he has developed a clear understanding and involvement regarding the “cure”, as he calls it.
He returns to school, to his work and to his mates, willingly. The night appointment with the “machine” and with its “medical treatment” has become part of his daily routine, so much that he falls asleep serenely well before I connect him to the dialysis machine. Often he wakes up crying, as before, but he never complains about this new thing he has to bear!
This experience, that made him grow, made us discover once more how marvellous and precious he is.
We are now hoping for a kidney transplant, knowing that this may also be a problem in our Country!
I have no news regarding other children in the world on dialysis because of Lesch-Nyhan and I ask myself: is it because kidney damage rarely becomes so serious?
- I have had news about a child with Lesch-Nyhan, who recently died of renal failure. -
Does the judgement of “impossible life” weigh on this difficult situation, too?
My thoughts wander; many considerations crowd my mind; then I look at Michele smiling at me and I see the certainty of the beauty of life; I taste the sureness of the value of *his* life.
One evening a father who lost his 14 year old boy, called me on the phone: he had got my number from the Association and had a lot to say about his lost “treasure”.
While he talks I feel with him and like him great emotion. I ask myself if these brave people who have had the experience of dealing with the illness, can make others more ignorant aware of how valuable the lives are of our special children. If we succeed in this, then perhaps **the “scientific” world will not find so “alien” to the need to fight to the end: the need to fight for the life and to better the quality**

of their lives.

CHAPTER 9

Growing up...

While these few pages were being written, our family moved towards a new experience: the meeting with a 35 year old man with the Lesch-Nyhan disease and the telephone contact with other 16,18, 28 year olds.
Once again we ascertained that the impression got from one of the first news letters, and quite recently too, about the illness, was wrong. That is the idea that the life expectation of these patients was limited to just a few years.
We now had proof that this “impossible life” is able to “resist” and surpass the trials of weariness, of the strain of the daily fight and of such an odd and cruel disease.

An exchange of smiles

Paolo, in his wheelchair in front of a padded table, with his wrists tied up with tape which limit his movements, received us with his face lit up by a really cordial smile.
Michele who, at home, had expressed a sort of dread for this meeting with an “elder man”, immediately answered back with his own cheerful smile.
We had interrupted a game of cards between our new friend and a pleasant lady careworker who several times a week fills up his time.
But Paolo did not seem to mind. He was looking at Michele with curiosity, attention and sympathy.
I however was looking at Paolo with sympathy and emotion: his presence simply opened up new prospects to me.
I looked at his smile made wider by his lower diminished lip: he too, just like the other youngsters who I know about, has fought his battles not without serious defeats...
Looking at his parents, I thought about their personal battle.
I was struck by his serenity, his equilibrium, probably achieved with great difficulty.

The dialogue with his parents coincided with our experience: Paolo actively contributes in indicating the dangers for himself as well as for others, he is always attentive and participates fully in daily life. He would have less motor problems than he demonstrates, if he had not been disturbed by the impulse to self-injury.

I have not yet got over the emotion of this meeting. I have put it next to emotions I got from talking to the mother of the 18 year old boy, who goes to high school and is a painter and from the phone call with the careworker of a co-operative attended by the 28 year old young man: all experiences that make me look at Michele with a new light and with more pressing questions.

Knowing that life, in spite of everything, continues and that for these children the experience of “growing up” is possible, an intervention directed more to improve the quality of their life is made urgent.

It is true that exchange of experiences between families is already a possible help, but I also believe that it is our right even if the disease is so rare, to be able to find **qualified** people who can help us to plan the path to a greater autonomy and to develop our children’s capabilities.

Their situation is unusual. It is rare. For this reason, not all methods generally applied to children with spasticity or behaviour problems prove to be right for them.

Like therapy, rehabilitation needs new researches, studies and comparisons to gain further knowledge.

Of course, to dream about refresher courses for careworkers who take care of these patients (put all together, they add up to quite a number!) that would became springing boards to new strategies and new ways, may be naive. Deep in my heart, when I look at my and other children, I know that their right to a dignified, serene life cannot be denied because they are “different“. This difference must not become a handicap in the handicap of being affected by a **rare disease**. Growing up is not just a privilege granted by mother nature, but a conquest that we make all together.

Their progress is a sure sign of our progress .

After the news regarding the existence of young men became

known amongst parents with the initial little group of Lesch-Nyhan children, I reflected a great deal on the reaction of a mother who beside her joy of the possibility of a longer life expectation for her child, revealed to me her new anxiety: how will we manage them when we grow older?
If the problem of the future is always a big question for the parents of disabled children who are not self sufficient, then for parents of children with Lesch-Nyhan disease it can become a nightmare.
If things remain as described up to now, the risk of not finding adequate support for the problems of our sons is not so remote...
The urgency to fight for an improvement in conditions of our children's health (finding therapies that allow them to increase their capabilities, no longer hindered by behaviour that most people do not understand) and for different awareness of their "uncommon" exigencies cannot but push us out from isolation. Let's look for each other, family to family, and let's unite and find a road together so that these "possible" lives l may really be sustained.
Andrea, the 18 year old boy who paints has given Michele a gift: a painting titled HOPE. Now this painting hangs on the wall in Michele's room and it means both for him and for us just what his title says: the hope for a future in the future.

Diego's hope

Angela, the mother of Diego a 17 year old young man, confirms, how the urgency of a "special" support for the families is true and how research for very special rehabilitation is necessary.
Her words, and her recollections are very precious for me. I wish just to underline the aspect of the disease, that afflict many families, of which I have little experience. The strange form of self-injury that causes these boys, so sensitive and in most cases delicate, to have the impulse of verbal aggression, coarse language and bad behaviour, the consequence of which is to inflicting on themselves psychological suffering.
Here is an example of this typical behaviour:

..." I remember a Christmas dinner. The guests were all dear people. Diego was chatting like everybody, but he was paying attention to Maurizio, his best friend. Maurizio is a serious, reflective and patient old man who has been helping Diego since he was a child.
Diego, during the chat, begun to insult him and spat at him insistently.
As we saw what he was doing, we changed our seats, without attaching any importance to what was happening.
During the dinner the atmosphere became lively. My husband and I lost sight of Diego who continued in his behaviour towards Maurizio up to exasperation.
The final reaction was explosive and unexpected: Maurizio, red with anger, threatened Diego affirming that he would not see him anymore.
Diego became serious and bowed his head. We left the party immediately. I cannot describe our reaction: we felt desperate of what had happened, we had lost a precious friend.
In a short time things came out right. Maurizio was close to us again.
We know Diego well, but we are never prepared enough to precede and face his reactions.
Nobody would believe that Diego regrets and weeps desperately after having insulted somebody he loves and that he can be a cheerful companion for his schoolmates and for our family."...

I find very interesting too, the events linked to learning:
..."With regards to school I can say that the greatest problems are to be referred to the teachers. In fact Diego learns well, but his behaviour leads to misunderstanding.
I remember that one day he returned home from school in tears. The assistant told me the whole story: Diego was in the classroom during the Italian lesson. The supporting teacher was not there because she had been called by the Director. The Italian teacher was speaking and during a pause, Diego swore aloud, in the silence. All the students burst into laughers.
The Italian teacher, very angry, sent him out of the door, in the

passage.
The second year of elementary school and the third year of intermediate school were the worst because those supporting teachers did not understand him.
After one year of incomprehension and spites, here is the final exam. Diego is calm although he knows that he must pass a hard test. All the teachers are waiting for him in the classroom. He greets us with a reassuring smile and lets himself be led in the class.
We, the parents, look at each other silently and anxiously. We wait. At first two teachers come out the room. They seem to be in good humour, they even laugh. As they realised we had questioning glance they reassured us: " He made it".
The History teacher is radiant: "We all know who Mussolini was and what he has done, but only Diego - with a four letter word - succeeded in rendering the idea".
And also thanks to Mussolini, Diego got the diploma of "Scuola Media".
Now our boy attends the fourth year of secondary school, he uses the computer,is well integrated and has a lot of friends.
I report what his teacher has said: "I was very struck, last year, by Diego when I was doing a lesson on the "Divine Comedy". He was enchanted, with wide open eyes and with bated breath. Then, when I tested him, he showed to have understood well the essential concepts.
This year, for the first time, he has asked, once or twice, to read the texts by himself. He reads with difficulty, but he gets great satisfaction from it".
His classmates and the boys from other classes, after being at first a little bewildered, have begun to call him and to bring him in class with them, showing to appreciate his presence".

Angela concludes:
"Diego does not have his teeth anymore. He has his arms in splints. Everything around him is padded. He underwent plastic surgery on his nose. He wears a glove to defend one hand from the other. Nevertheless I can say that he has never lost his will to live and his hope to recover. Almost everyday he says: - If I could walk...I would do...I would go...I would

study for...etc. -“
Diego’s hope is our hope, too.
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CHAPTER 10

Remarks

The discovery of new energies in our “daily struggle” has in any case given a horizon of hope for the life of our children. It has rendered the desire to resist and fight even more “possible”.
Being acquainted with the hundreds of cases of rare syndromes has made me realise how “common” the situation of being affected by a rare disease. This fact has spurred on new considerations. It is true, Michele is one of the **few** boys with Lesch-Nyhan Syndrome, but he is one of the **many** whose life is considered “impossible” because of the unusual and varied characteristics of rare diseases.
Therefore maybe it is quite *normal* and *common* that Medicine has still many frontiers to cross, a long way to go because there are so many different diseases, but the battle against all forms of sickness and death is part of the *possible* life.
Of course a surrender is possible, but throughout history, man has shown a force with which to overcome obstacles.
He has pushed himself beyond the barriers and has raisen again and again from his defeats.
Maybe this force is present in the very “genetic code” of humanity...
“For love is as strong as death” says the Song of Songs.
Each one of us can name this vital energy as he wants, but certainly, without it, the life of *each man* becomes really

impossible.

PAOLA
Michele’s mum.

Dedicated to Michele

I think it is significant to present to you a poem that little Michele has inspired a twenty year old young poet Marco Guidetti to write:

Stairs to moon

Your five years of winged thoughts
fly closer and closer to the reefs
of outstretched hands, who, each day
elbow just to caress your breath.
From the images that windows
rudely yawn
you always look for the shrillest note of the sun,
even when the adagio of the night
absentmindedly strolls on world's back.
The host of souls that continuously
gravitates round your nest,
savours,
for the definitive today's last time,
the freshness of trees
who fight against
a sharp January dawn wind,
the dense heat of last October sea,
the dazzling smile
of a first June morning.
Loneliness is a jail in the open air,
the death sentence more voted by civil society.
But you will never be a prisoner in freedom.
The moon may seem so far to you,
but if you climb up the stairs
of my emotions,
you will not get tired reaching it.
Your mother trusts,
your father dreams.
Friends stop the clouds in
a hurricane night to get noticed

by your perpetually astounded expression.
You would say thanks to everybody,
but the well in the bottom of which
the alphabet invites you
is too deep to dive
barefoot.
You have not been surrendering
for this reason
and you roll your crazy pupils,
as if they slide on a frozen bend,
you open your arms
as if you had to hold to your chest
the entire world.
But your hands are two butterflies in a larva,
so small they seem invisible,
so delicate you can only stroke them.
Whoever stands in front of you
he hates you for the way you show your life
he envies you for each clove of day
sipped as water in the desert,
he loves you because he would not change
a sigh of your human being.
This time I say thanks to you.

Marco Guidetti.