Istituto di Clinica Pediatrica
Cattedra di Pediatria
Università Cattolica Sacro Cuore

Maratona di :
Esperienze di Assistenza Integrata al Bambino con Malattia Rara e alla Sua Famiglia

5 Luglio 2002

Observing Simona's growth for 14 years, del 5p-, and that of her family: the pediatrician narrates:

Flavia Luchino
Family pediatrician

(water in the background, Simona's favorite element)

an afternoon 14 years ago

- two very young parents placed

  a 2-month-old baby and a cytogenetic report on the examining table of my office:
- Del 5p-
- I was aware that they were observing my facial expression in hopes of gleaning information.
- and **I had no information**

1:50,000. And here she is: Simona

our only source of information:
the parents tell me about Simona’s likes and dislikes
how she reacted or didn’t react
to cuddling or discomfort:

they had already met Susanna,
the physiotherapist of the ASL (local health unit), to whom they had been referred by the genetics center, who in turn sent them to me.

searching for information

- The textbooks were full of stereotypes
- The genetics department, who remembered another case whom they lost touch with, offered a yearly check-up at their Day Hospital
- Internet didn’t exist yet (it was the year 1988)
- The children with Cri du Chat association was founded in 1995
- At the library...

at the library there were only two articles

people who were raised by their families,

retrospective evaluations:

serious delays, **with significant variation** in motor,cognitive, behavioral and language development

- they walk between 2 and 8 years of age, manifest autistic behavior,express themselves through sounds or gestures which are comprehensible to their relatives

Wilkins LE, Brown JA, Wolf B. Psychomotor development in **65** home reared children with cri-du-chat syndrome.J Pediatr. 1980 Sep;97(3):401-5.

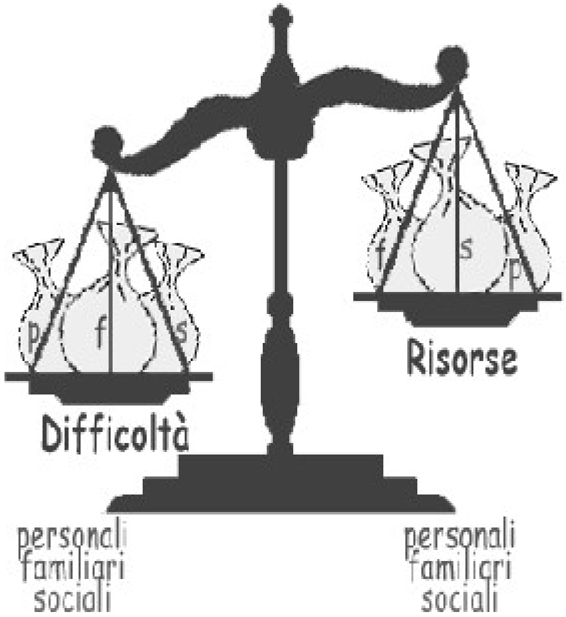
Wilkins LE, Brown JA, Nance WE, Wolf B. Clinical heterogeneity in **80** home-reared children with cri du chat syndrome.J Pediatr. 1983 Apr;102(4):528-33.

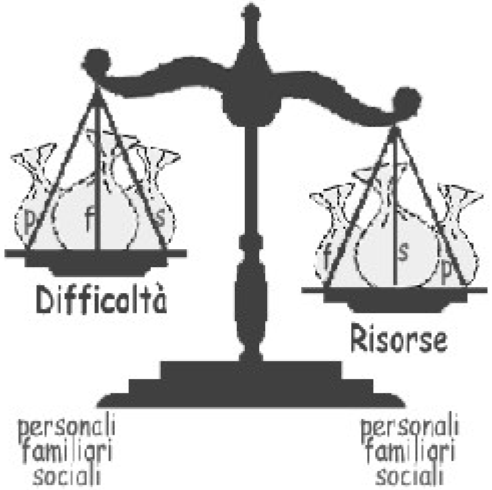
significant variation...?

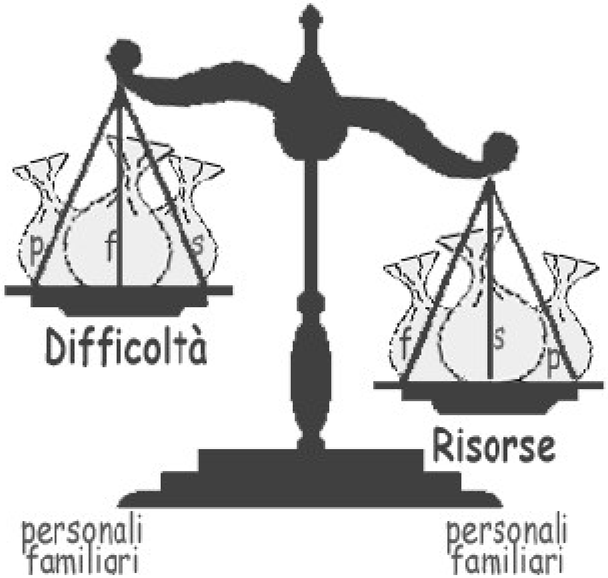
we decided to look for a guide
just in Simona,
her parents
and in a conceptual framework
of vital concepts,
shared during years
of multidisciplinary work

a conceptual framework of vital concepts shared with the family too

- the uniqueness of development, its observation
- the importance of a global view of the child
- the influence of the family environment on the child's development
- the home visit as the basis for support and observation
- the participation of the family in decisions which affect them
- the inadequacy or the danger of rigidly applied methods
- the multifactorial assessment of difficulties and individual, personal and social resources







the multidisciplinary team
met with the family

and periodically weighed
difficulties and resources on a conceptual scale.

initial difficulties

the scale tipped violently towards needs:

- pain
- lack of information
- fear of inadequacy
- a sense of isolation
- sluggishness of a child who doesn’t respond
- hypotonia which complicates feeding too

the initial resources

ne family, the ASL (Local Health Unit), the pediatrician, the daycare center:

the home visits, the pediatricians and healthcare professionals

psychological help for the parents

the meetings with the grandparents

social work

the physiotherapist’s support of her development

gradual inclusion at the daycare center

being intersupportive:

the only information available was that which we were able to gather with the parents, every day, which had to be shared amongst us.

yearly check-up at their Day Hospital of Genetic Department...

ay Hospital presso il Servizio di Genetica Medica ,
spedale pediatrico B. Gesù. Roma, 16/03 1990

i Genitori e p.c. al Loro Medico Curante

a visita neurologica ha riscontrato un miglioramento della partecipazione ambientale e dell’organizzazione motoria rispetto al precedente controllo. Si consiglia di proseguire con la riabilitazione impostata.

a visita cardiologica non ha evidenziato rilievi patologici. L’esame ECG e' nei limiti della norma per l’eta'.

el corso del Day Hospital sono stati eseguiti alcuni esami di laboratorio: emocromo, glicemia, azotemia, creatinina, proteine totali, colesterolo, trigliceridi, calcio, fosforo, fosfatasi alcalina, transaminasi, gammaGT, LDH, CPK ed immunita’ cellulare sono risultati nella norma. L’immunodiffusione ha rilevato valori normali di IgG e IgA associati a valori di IgM superiori alla norma (326 mg% - V.N. 36/104).

estiamo a disposizione per ogni eventuale chiarimento (tel. 06/65192227), consigliando inoltre un controllo in Day Hospital presso il Servizio di Genetica Medica tra 1 anno.

even small unforseen difficulties came up:

for example, transportation to the daycare center, or a courageous babysitter or feeding and personal care assistant (changing diapers) at school couldn’t be found,

which in turn **activated**, when needed,

more social, family and personal resources:

volunteers, grandparents, mother who gets her driver’s license, healthcare workers who donated their free time…

but the family carried the true, unrelenting daily load

the parents’ words:

“ For a parent, a “special” daughter is someone to protect, it seems like anything could happen to her, the difficulties initially were great; from feeding her, to attempts at getting her attention, to understanding her smile…a facial expression”

the difficulties parents of “special” children encounter

are always with the various state and local institutions:

at each request it seems as though

you are stealing,

there is no personal contact

you are only a “case”

the parents speak further,

the true great directors:

all the healthcare workers took our Simona to heart and supported us in choices to come.

In the schools the difficulties came from the school administration but the teachers were always by our side.

A great help came from the home visitor, Maresa, who succeeded in forming a relationship based on friendship and respect with Simona.

with Federica’s birth,

two years later, we feel like we have twins.
Simona is constantly stimulated
by her presence,
and when Chiara is born 8 years later,
Simona understands
that she is not the baby of the family
n the meantime, Federica becomes a young lady
and wins singing contests…
Chiara grows up content and curious.

an important moment: ABC (the Cri du Chat Association)

a great resource was when

the Cri du Chat Association was born:

gatherings of scores of families with their children, scientific research, conferences as of 1995.

a big emotion

the end of isolation

the availability of information

The Cri du Chat Children’sAssociation

Tel: 055 828683 e-mail:abc.fi@tin.it

Recent studies on cognitive levels and behavioural disorders, 5p-:

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Genotype-phenotype correlation :a progressive severity of clinical manifestation and psychomotor retardation related to the size of the deletion.

REGIONE PIEMONTE, OSPEDALE S. ANDREA.VERCELLI
DIVISIONE DI PEDIATRIA E PATOLOGIA NEONATALE,
SERVIZIO DI GENETICA

Vercelli, 11/12/01

- L’esame effettuato a Simona con le tecniche di citogenetica molecolare conferma la presenza della delezione di 5p e dimostra che sono state perdute le "regioni critiche“ per pianto, ritardo cognitivo medio-grave e microcefalia. La più precisa definizione dell’ampiezza della delezione, ottenuta con la tecnica utilizzata, ha consentito di dimostrare che il punto di rottura è a livello della banda p13 e compreso tra gli YAC 921F7 e Y816F10 (Fig. 2).

Premesso che altri fattori genetici o ambientali possono influenzare il fenotipo, la valutazione clinica (compatibilmente con i dati disponibili) insieme alla definizione citogenetico-molecolare fanno ritenere che Simona presenti una forma di media gravità, grazie anche agli interventi riabilitativi ed educativi della famiglia e di tutti gli operatori.

Prof.ssa Paola Cerruti Mainardi

E. O. Ospedali Galliera di Genova-Laboratorio di Genetica Umana

Indagine citogenetica molecolare

molecular cytogenetic analysis

Data: 17.04.2001

nviato da: Prof. P. Cerruti Mainardi

otivazione FISH: Analisi struttura braccio corto del cromosoma 5.

essuto esaminato: Metafasi di linfociti di sangue periferico.

onde utilizzate: MEGA YAC del CEPH: 767E1 (D5S471,405,417,675)921F7(D5S661),816F10(D5S651,674)

ilevazione e controcolorazione: AVIDINA - CY3, DAPI

ellule in metafase esaminate: Totale n° 30

ARIOTIPO COSTITUZIONALE: 46XX,ishdel(5)(pter p13)(Y921F7 -)

onclusioni: L’ibridazione in situ eseguita con le sonde sopracitate ha
videnziato che il punto di rottura della delezione del braccio corto è a
vello della banda p13 ed è situata tra gli YAC 921F7 e Y816F10.

Simona today

A Brunet Lezine evaluation performed 2 years ago, showed a developmental age between 18-24 months.

But **Simona astonishes us because she doesn’t fit a model,** she changes every day, she learns in slow motion, and she remembers, showing a better receptive than expressive language skills.

She has her interior world, of 14 years of a full life, led in her own special way. She expresses herself with just a few words or sounds, but sits well at the table and listens, she helps more and more every day in self-care skills, she respects the rules, expresses feelings, adores water, and swims carefree and calm.

Simona next year

a work in progress:

focusing on objectives

a complete rehabilitation plan

the objectives :

including her in the new middle school: reciprocal adaptation

independence in basic self-care skills

communication and affects

behavior and rules

goal-oriented tasks to occupy her time

sports (horse-back-riding and swimming)

preventing excessive weight gain, cavities and scoliosis

the family’s well-being and the sisters’ right to grow up free and independent

the complete rehabilitation plan,

that involves new and old actors:

- the growing family (Federica in middle school, Chiara in elementary school)
- Simona’s new middle school
- the family pediatrician
- the healthcare workers at the (ASL) local health unit
- the SAISH (after school care at home)
- the “Casetta” (a play space , law number 285, 1997, for the afternoon, and some weekends)
- speech therapy a home, coverered by the National Healthcare Plan
- sports (horse-back-riding and swimming paid for by the family)

and when Simona grows up?

In the local health unit’s (ASL RME) territory,

a service for residents over 18 years of age, who have a confirmed and stable diagnosis,

is available (run by a multidisciplinary healthcare and social work team).

In the last two years, the following have been opened:

3 day centers (80 consumers)

9 “foster”residential care homes (case famiglia) and residential communities in collaboration with the Ciy of Rome

(63 residents)

1 residence for patients with complex pathologies (20 persons)

Day centers for adults

The following items are predicted:

from law number 104,1992,

rehabilitation guidelines of the Health Ministry,1998,

from the law number 162, 1998,

from the National Health Plan

from the Regional Health Plan

This summer Simona will go

to summer camp, 15 days. Day and night

(financed by the Local Health Unit (ASL) and the City of Rome)

She has already started to sleep alone at her house or at the “Casetta”, with the assistants she knows best, while Mom and Dad take Federica out of town for a singing contest, or just to have a vacation with Chiara too.

And in the future
we still want to be united

because one day Santina,
Simona, Federica and Chiara's mother,

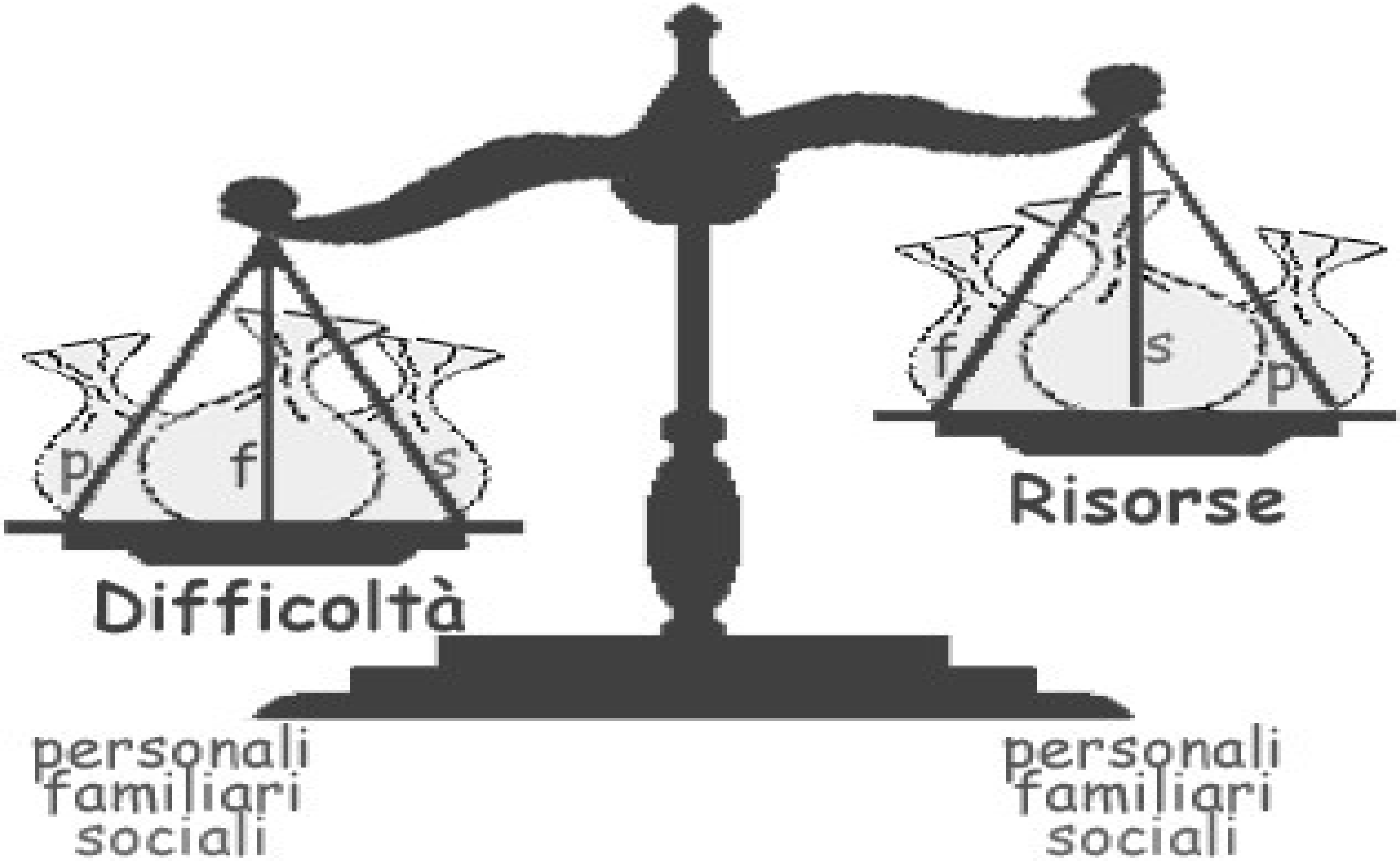
can hear society tell her,
as the home assistant often did:

“Santina,

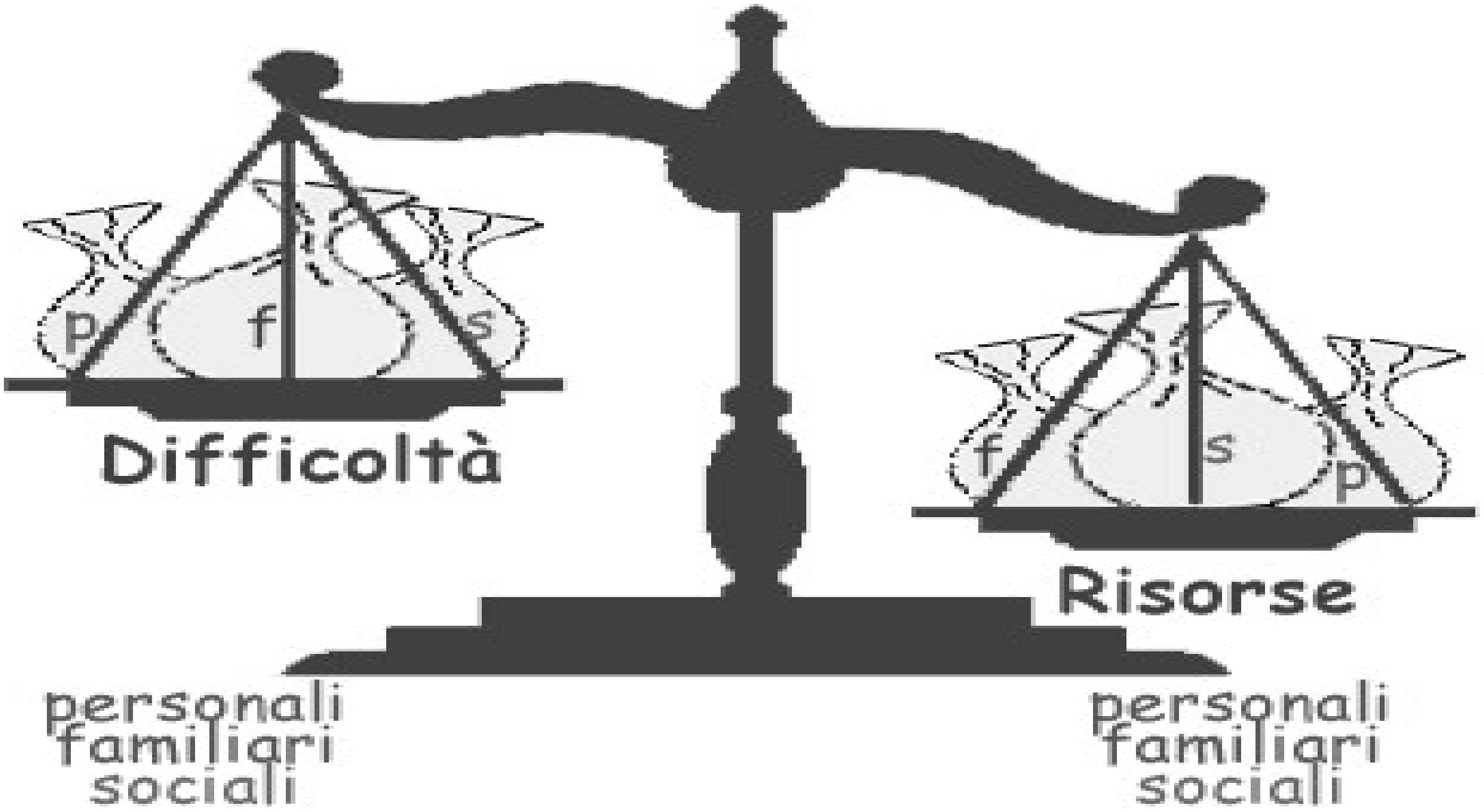
I’ll take care of things now,

you go and rest”.

Weighing difficulties and resources on the scale: personal, family and social



Weighing difficulties and resources on the scale: personal, family and social



And which resources are available for a family pediatrician in the ‘80’s? Thanks to all those who guided me towards observation and respect

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http://www.aap.org/policy/pprgtoc.cfm

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SN 2002-2004. Tra i 10 obiettivi strategici:Creare una rete integrata di servizi sanitari e sociali per l’assistenza ai malati cronici, agli anziani e ai disabili

The APeC’ s proposal
(Community Pediatric Association)

This article takes the Italian socio-legislative situation into account and acts as a guide at a local level, for those who have to organize managed care for chilren who are affected by chronic diseases.

ApeC- (Associazione Pediatria di Comunità- Association affiliated with the ACP, Pediatrician’s Cultural Association)

Assistance for children and teens with chronic diseases

Quaderni acp 2002; vol IX n. 1:56-58

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abstract

The lack of sources of information most certainly affected the beginning of this story.

In a negative sense, for obvious reasons,but also in a positive sense, because it activated the same number of resources that could be found in a well-organized “individual assistance plan” today.

f back then everything was tied to the unpredictable and spontaneous unity of health care workers that shared the same goals, now with the possibility of transferring and multiplying information, and the new approach of the National Health Plan, it is possible to follow similar paths more effectively.

The focus of the public administration can and must now converge more incisively on the enormous

open-ended problem of adult’s life of our children.

in collaboration with:

- **Flavia Luchino, pediatra di famiglia,**
- **Santina e Bruno Graziani,**

  Simona’s parents
- **Federica e Chiara Graziani,**

  Simona’s sisters
- **Maresa Vanni,assistente domiciliare SAISH MunicipioXX**
- **Nadia Russo, assistente domiciliare SAISH MunicipioXX**
- **Susanna Schroder, fisioterapista ASLRME**
- **Maria Vittoria Leonardi, fisioterapista ASLRME**
- **Elena von Eles, assistente sociale ASLRME**

- **Cosimo Baldari, Educatore U.O.S.E.C.S. MunicipioXX**
- **Enrico Nonnis, neuropsichiatra**

  ASL RME
- **Nora Ferro, psicologa ASLRME**
- **Paola D'Alessio, psicologa. ASLRME**
- **Rina Fontanelli, psicologa ASLRME**
- **Nelvia Costa, psicopedagogista Scuola Materna Statale**
- **Cira Somma,insegnante di sostegno Scuola Materna Statale**
- **Maria Elena Alberti,insegnante di sostegno Scuola Elementare Statale**
- **Franca Minnocci, insegnante di sostegno Scuola Elementare Statale**
- **Laura Piccinino, Educatore ASL RME (Area Disabilità Stabilizzata)**

Maratona di : Esperienze di Assistenza Integrata al Bambino con Malattia Rara e alla Sua Famiglia

Roma, 5 Luglio 2002

Finally, a bridge! A bridge between numbers and life.

Epidemiology and molecular biology, genetics: they are up there counting with a goal of prevention. Indispensable, it is true. But assistance, life, and its quality, for those who in the data banks are just a number with a positive sign, (I mean those who eluded prevention, and are useful only for frequency histograms)are down here fighting and suffering, without any means of being heard.

Who knows if this idea is only utopia , but one which would allow us to make a dream of hope come true. The hope of helping one another to live and help others.

I want to believe in this dream: respectful of the people with whom we shared so many years of hard work and hope.