

The quality of life in girls with Rett syndrome

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Abstract

Nowadays, quality of life is receiving an increasing attention in all scientific areas. Rett syndrome (RTT) is a rare neurological development, affecting mainly females. The congenital disease affects the central nervous system, and is one of the most common causes of severe intellectual disability. The aim of our study is to evaluate the effect of RTT on the quality of life of people who are affected. Both parents of 18 subjects, all female, diagnosed with RTT, took part in the research. Quality of life was assessed using the Italian version of the *Impact of Childhood Illness Scale*. This scale consists of 30 questions that investigate the effect of illness on children, parents and families. For each question, the parent was asked to rate two variables: frequency and importance. Another questionnaire was administered to obtain medical history, diagnostic and therapeutic data of the persons with RTT. Our data show that RTT has a considerable impact on both the child's development and the entire family. Parents' answers demonstrated that their child's illness had consequences for the child and how the family coped with it. For this reason, attention should be directed at psychological and social aspects, as well as attitudes, manners, reactions and effects such disturbances can have on the entire family.

Introduction

Rett syndrome (RTT) is a progressive neurodevelopmental disorder that tends to affect mainly girls during the first years of life, after a period of apparent normality: it is a disease that is the second leading cause of intellectual disability in girls, with an estimated incidence of one in 10,000 females born.¹⁻⁵ After seemingly normal development in the first/second year of life, the disease is manifested by a gradual loss of language and manual skills, accompanied by hand stereotypies, acquired microcephaly, growth slowdown, epilepsy, ataxia, respiratory disorders (hyperventilation and apneas). Sleep disturbances and increase of total sleep time or numerous night awaken-

ings can be also showed.⁴⁻⁸ Epilepsy affects the majority of patients in a specific clinical stage of the disease and it is drug resistant in approximately one-third of cases.⁸ For years, the RTT has been incorrectly classified as autistic disorder.

The disease has genetic basis and it is determined by the mutation of a gene, *MECP2*, on the X chromosome, which encodes the methyl-CpG-binding protein 2.⁹⁻¹¹ There are variants of the syndrome with symptoms attenuated, anticipated or deferred, or with the absence of one or more typical symptoms.¹² These different clinical forms are due to the complex interaction of different parameters, such as the type of mutation in the *MECP2* gene, its location in the functional domain of the gene, the existence of a total or partial inactivation of the X chromosome carrying the mutated gene.⁹⁻¹¹ There would be other genes involved in the modulation of the *MECP2* gene that may explain those cases of RTT in which was not identified any mutations in *MECP2*.⁹⁻¹¹ It was also accused another gene called *CDKL5*.⁹⁻¹¹ The association of epilepsy and even drug-resistant epilepsy has been reported in certain genotypes of the methyl-CpG-binding protein 2 mutation, which is present in a majority of patients with classical RTT.⁸

The aim of our study is to evaluate the effect of RTT on the quality of life of people who are affected and on their parents.

The effect of RTT on the quality of life of affected girls' families was assessed using the *Impact of Childhood Illness Scale*.¹³ This questionnaire assesses the quality of life in children with epilepsy and other chronic pathologies and in their families, as well as in other studies.¹⁴⁻¹⁶

Materials and Methods

The study includes parents of 18 girls with RTT, residing in various Sicilian provinces. They were recruited from the Italian Association of Rett (AIRETT). All girls had sleep disorders and seizures. Girls were an age between 10 and 16 years, fathers ranged in age from 30 to 57 years, while the mothers from 28 to 52. The parents' education level (higher among the fathers than the mother) was middle-high: secondary school (21 parents), middle school (8 parents), University (7 parents).

The effect of RTT on the quality of life of families of affected girls was assessed using the *Impact of Childhood Illness Scale*.¹³ All questions refer to the effect of the illness on the child, the parents and the whole family. The scale comprises 30 questions divided into four sections: impact of illness and its treatment (questions 1-5); impact on development and child's adjustment (questions 6-15);

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impacts on parents (questions 16-20) and impacts on the family (questions 21-30).

For each question, the parent was asked to rate two variables: frequency and importance. The first refers to how often a particular problem or situation arose (never or rarely true, sometimes true, often or really true) and the second to the amount of concern it produced (not much concern, a bit of concern, a lot of concern). The two variables for each question were scored 0.1 or 2. The questionnaire also contains a brief preliminary section that explains to parents the aim of the assessment procedure.

Moreover, during the interview with parents, demographic data and information about the child's illness have been collected: child's age, pregnancy and birth, psychomotor development stages, age at which the child started school, subject's acquired and reached abilities, age at diagnosis, possible associated disturbances or pathologies, diagnostic examinations (electroencephalography, magnetic resonance imaging), drugs administered and educational programs. Items on psychopharmacological or psychotherapeutic treatment for one both parents could also be included. Descriptive analysis was used for collected data, by means of parameters of frequency and importance for each question.

Results

The results of the questionnaire administered to parents of girls affected with RTT are

showed in Tables 1-4. The most recurrent problems connected to the child's illness, derived from an analysis of frequencies and percentages of answers for both parents (who answered *often or really true* according to the frequency parameter and *a lot of concern* according to the importance parameter) were for questions 18, 20, 21 and 13: self-care skills (frequency of fathers = 15 subjects; frequency of mothers = 12; importance of fathers = 11; importance of mothers = 12), difficulty in explaining the child's illness to her (frequency of fathers = 8 subjects; frequency of mothers = 11; importance of fathers = 13; importance of mothers = 15), difficulty in giving necessary attention to the other children in the family (frequency of fathers = 14 subjects; frequency of mothers = 16; importance of fathers = 13; importance of mothers = 13), difficulty to find a job for child after the school (frequency of fathers = 9 subjects; frequency of mothers = 10; importance of fathers = 11; importance of mothers = 10).

Vice versa, on questions 1 and 3, the majority of parents answered *never or rarely true* according to the frequency parameter and *not much concern* according to importance. In particular, they did not consider as complications of RTT disease the possibility that children may stop breathing (frequency of fathers = 16 subjects; frequency of mothers = 17; importance of fathers = 16; importance of mothers = 18) and that illness may damage their brain or die (frequency of fathers = 14 subjects; frequency of mothers = 15; importance of fathers = 15; importance of mothers = 16).

Finally, another interesting fact is that both parents declared that the child's illness did not affect their job opportunities (question 30): frequency of fathers = 12 subjects; frequency of mothers = 15; importance of fathers = 19; importance of mothers = 11.

Discussion

RTT remarkably influence the life of those affected and their families. For this reason it is important and necessary to direct attention not only to the kind of the disorder, but also the impact the disorder has on the child, parents, consanguineous and the entire family as a whole. Studies using the Impact of Childhood Illness Scale to investigate the effect of a child's chronic and invalidating pathology on his/her parents have pointed out that these illnesses can influence not only the child's development, but also the life of the entire family.¹³⁻¹⁵ Previous researcher on children with RTT has focused primarily on the causes and treatment of the illness, whereas our study is one of the few in which both parents were interviewed.

The parents recognized their difficulties in explaining their child's illness to others and to the child herself, probably because of the psychosocial implications of the illness.^{14,15} They

Table 1. Answers of fathers on frequency.

Questions	Answers (No.)		
	Never or rarely true	Sometimes true	Often or really true
1) Because of his/her illness he/she may stop breathing	16	0	2
2) There is a risk he/she may injure himself/herself	4	10	4
3) There is a risk he/she may be brain damaged or even die	14	3	1
4) The drugs my child takes makes him/her less alert	15	2	1
5) The drugs worsen his/her behavior	4	11	2
6) My child is more moody because of his/her illness	7	8	3
7) He/she is shy and more easily embarrassed	8	7	3
8) Because of his/her illness, he/she is teased and bullied	5	11	2
9) Because of his/her illness, he/she has few friends	4	8	6
10) Because of his/her illness, he/she has fewer interests	3	7	8
11) Because of his/her illness, my child has problems in reading or math	3	11	4
12) My child is less clever because of his/her illness	3	9	6
13) My child may not find a job when he/she leaves school	3	6	9
14) My child may have to take drugs for years	5	10	3
15) My child may not marry or have a family	3	8	7
16) My child makes a fuss about taking his/her drugs	15	1	2
17) Because of his/her illness, it difficult for him/her to use public transport	4	5	9
18) He/she is less able to care for him/herself	1	2	15
19) It is difficult to explain my child's illness to others	2	15	1
20) It is difficult to explain my child his/her own illness	2	8	8
21) Because of his/her illness, my child must be more closely watched than other children	1	3	14
22) It is difficult to give my other children enough attention	3	13	2
23) My child illness limits what his /her brothers and sisters can do	4	13	1
24) We have to limit our holidays	8	9	1
25) His/her illness means we have fewer friends around	10	5	3
26) My child illness influence how often we go out as a family	11	7	1
27) We have more arguments at home	16	1	1
28) We go out less often in the evenings as a couple	11	5	2
29) My child is more difficult to manage because of his/her illness	1	9	8
30) Because of his/her illness we turn down opportunities at work	12	4	2

believed that the psychological, social and practical difficulties of living with a RTT-affected girl are not completely understood by others.¹⁷⁻¹⁹ Therefore, operators, doctors, psychologists, educators who treat these persons should pay particular attention also to their psychosocial spheres. This requires to understand the interior life of the child and the psychological interactions with their illness, and involves the affected child's parents and consanguineous as well. Previous research on children with RTT has focused primarily on the causes and treatment of the illness, whereas our study is one of the few in which both parents were interviewed.¹⁷⁻¹⁹

Similar studies have been conducted with the same questionnaire to parents of children with pervasive developmental disorder, children with chronic kidney disease and others with epilepsy.¹³⁻¹⁵ These studies showed the same significant impact of the disease on parents, showing no differences from the present study.

Conclusions

The girls with RTT have a stop in the development followed by a psychomotor regression. They progressively lose the skills previously acquired, as the purposeful use of the hands, the language, the deambulation and communication skills and, moreover, autistic traits appear. They also begin to manifest seizures, cardiac and breathing problems. Nowadays, pharmacological therapy is essentially symptomatic and aimed at treating symptoms such as seizures, sleep and behavior disorders.⁴

In order to improve their quality of life is necessary that parents give a meaning to the behaviors emitted by the girls: some of them may occur out of habit or as an outburst of a stress, others might have communicative value.^{4,7}

The parents will manage the problematic behaviors by implementing cognitive-behavioral techniques.^{4,7} To determine the reason for which a girl with RTT is manifesting a certain behavior it is recommended to consider

her physical state, the situation in which the behavior occurs and have a good understanding of her usual concerns.^{4,7}

All modalities of intervention must be supported by an action on the living environment of the girls. Parents should be in harmony with the needs of girls and be able to read their signals.^{4,7}

To this end, a particularly useful measure is the restructuring of the environment and to resize parental expectations within the abilities of girls, using a positive approach in re-education, in order to decrease the level of anxiety and increase the self-esteem.⁷

All this will lead to a better quality of life for individuals with RTT.

This study compares only some aspects of the quality of life of subjects with RTT, limiting the focus to some areas to be further developed in more extensive studies. These subjects, for example, can improve their quality of life by means of the direct contact with nature and enjoyment of the rural landscape thanks also to the multifunctional role of farms.²⁰⁻²²

In other studies has been noted that a

Table 2. Answers of mothers on frequency.

Questions	Answers (No.)		
	Never or rarely true	Sometimes true	Often or really true
1) Because of his/her illness he/she may stop breathing	17	1	0
2) There is a risk he/she may injure himself/herself	4	11	3
3) There is a risk he/she may be brain damaged or even die	15	2	1
4) The drugs my child takes makes him/her less alert	16	2	0
5) The drugs worsen his/her behavior	17	1	0
6) My child is more moody because of his/her illness	10	6	2
7) He/she is shy and more easily embarrassed	10	7	1
8) Because of his/her illness, he/she is teased and bullied	11	7	0
9) Because of his/her illness, he/she has few friends	2	6	10
10) Because of his/her illness, he/she has fewer interests	1	7	10
11) Because of his/her illness, my child has problems in reading or math	1	6	11
12) My child is less clever because of his/her illness	2	3	13
13) My child may not find a job when he/she leaves school	4	4	10
14) My child may have to take drugs for years	5	4	9
15) My child may not marry or have a family	2	5	11
16) My child makes a fuss about taking his/her drugs	11	6	1
17) Because of his/her illness, it difficult for him/her to use public transport	9	4	5
18) He/she is less able to care for him/herself	2	4	12
19) It is difficult to explain my child's illness to others	3	9	6
20) It is difficult to explain my child his/her own illness	2	5	11
21) Because of his/her illness, my child must be more closely watched than other children	1	1	16
22) It is difficult to give my other children enough attention	1	16	1
23) My child illness limits what his /her brothers and sisters can do	2	13	3
24) We have to limit our holidays	14	1	2
25) His/her illness means we have fewer friends around	8	7	2
26) My child illness influence how often we go out as a family	6	7	5
27) We have more arguments at home	9	6	2
28) We go out less often in the evenings as a couple	5	8	5
29) My child is more difficult to manage because of his/her illness	2	8	8
30) Because of his/her illness we turn down opportunities at work	15	2	1

severe clinical impairment was highly associated with poor physical quality of life, but worse motor function and earlier age at onset of RTT stereotypies were associated with better psychosocial quality of life; conversely, better motor function was associated with poorer psychosocial quality of life.²³

RTT have a notable impact on both the child's development and the whole family. Parents' answers point out that their children's illness influences their adaptation, worsening the clinical case history.^{13-16,24,25} For mothers of children with RTT syndrome the most important predictors of maternal physical and emotional health are child behavior, caregiver demands, and family function.²⁶ Moreover, according to other studies, mothers giving care to children with RTT are at high risk of severe depression.²⁷

So, the approach to a person with RTT should be global; in addition to the intervention on the person, an adequate psychological support to parents and family should be provided.^{14,15} It is important to underline how

numerous variables and their reciprocal interactions can affect children with RTT and the entire physical, psychological and social adaptation of family.^{13-15,17-19}

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Table 3. Answers of fathers on importance.

Questions	Answers (No.)		
	Never or rarely true	Sometimes true	Often or really true
1) Because of his/her illness he/she may stop breathing	16	2	0
2) There is a risk he/she may injure himself/herself	5	12	1
3) There is a risk he/she may be brain damaged or even die	15	3	0
4) The drugs my child takes makes him/her less alert	16	2	0
5) The drugs worsen his/her behavior	15	3	0
6) My child is more moody because of his/her illness	9	7	2
7) He/she is shy and more easily embarrassed	8	8	2
8) Because of his/her illness, he/she is teased and bullied	9	8	1
9) Because of his/her illness, he/she has few friends	4	9	5
10) Because of his/her illness, he/she has fewer interests	1	5	12
11) Because of his/her illness, my child has problems in reading or math	2	7	9
12) My child is less clever because of his/her illness	3	4	11
13) My child may not find a job when he/she leaves school	4	3	11
14) My child may have to take drugs for years	4	11	3
15) My child may not marry or have a family	4	4	11
16) My child makes a fuss about taking his/her drugs	13	4	1
17) Because of his/her illness, it difficult for him/her to use public transport	5	4	9
18) He/she is less able to care for him/herself	2	5	11
19) It is difficult to explain my child's illness to others	3	11	4
20) It is difficult to explain my child his/her own illness	2	3	13
21) Because of his/her illness, my child must be more closely watched than other children	1	4	13
22) It is difficult to give my other children enough attention	2	11	5
23) My child illness limits what his /her brothers and sisters can do	5	10	3
24) We have to limit our holidays	11	4	3
25) His/her illness means we have fewer friends around	3	12	3
26) My child illness influence how often we go out as a family	9	5	4
27) We have more arguments at home	8	4	4
28) We go out less often in the evenings as a couple	3	8	7
29) My child is more difficult to manage because of his/her illness	1	8	9
30) Because of his/her illness we turn down opportunities at work	9	7	2

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Table 4. Answers of mothers on importance.

Questions	Answers (No.)		
	Never or rarely true	Sometimes true	Often or really true
1) Because of his/her illness he/she may stop breathing	18	0	0
2) There is a risk he/she may injure himself/herself	4	13	1
3) There is a risk he/she may be brain damaged or even die	16	2	0
4) The drugs my child takes makes him/her less alert	17	1	0
5) The drugs worsen his/her behavior	15	3	0
6) My child is more moody because of his/her illness	12	5	1
7) He/she is shy and more easily embarrassed	9	8	1
8) Because of his/her illness, he/she is teased and bullied	12	6	0
9) Because of his/her illness, he/she has few friends	4	8	6
10) Because of his/her illness, he/she has fewer interests	1	4	13
11) Because of his/her illness, my child has problems in reading or math	1	8	9
12) My child is less clever because of his/her illness	3	4	11
13) My child may not find a job when he/she leaves school	4	4	10
14) My child may have to take drugs for years	4	11	3
15) My child may not marry or have a family	3	4	11
16) My child makes a fuss about taking his/her drugs	16	1	1
17) Because of his/her illness, it difficult for him/her to use public transport	4	4	10
18) He/she is less able to care for him/herself	2	4	12
19) It is difficult to explain my child's illness to others	2	12	4
20) It is difficult to explain my child his/her own illness	0	3	15
21) Because of his/her illness, my child must be more closely watched than other children	1	4	13
22) It is difficult to give my other children enough attention	1	3	14
23) My child illness limits what his /her brothers and sisters can do	4	11	3
24) We have to limit our holidays	3	8	7
25) His/her illness means we have fewer friends around	13	2	3
26) My child illness influence how often we go out as a family	4	10	4
27) We have more arguments at home	13	4	2
28) We go out less often in the evenings as a couple	3	10	5
29) My child is more difficult to manage because of his/her illness	1	8	9
30) Because of his/her illness we turn down opportunities at work	11	6	1
30) Because of his/her illness we turn down opportunities at work	9	7	2