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# THE QUALITY OF LIFE OF THE PARENTS OF A CHILD WITH RUBINSTEIN-TAYBI SYNDROME

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## SUMMARY

**Background:**

The goal of our study was to examine the functioning of a family with a child who has Rubenstein-Taybi syndrome (RSTS) in respect to the quality of life (QOL) of the child’s parents. We used measures associated with the parents’ QOL, an evaluation of the child’s psychomotor development and temperament, sociodemographic variables, and psychological variables, such as parental attitudes and coping styles.

**Material/ Methods:**

We used our own clinical history questionnaire; participatory observation; analysis of medical documentation; the Self-Anchoring Ladder; the Satisfaction with Life Scale; the Good Marriage Questionnaire; the Parental Attitudes Scale; the Coping Inventory for Stressful Situations; the Emotionality, Activity, and Shyness temperament questionnaire; and the Bayley Scales of Infant Development – Third Edition.

**Results:**

The child’s disability affects the parents’ QOL negatively. In this case, the child’s mother subjectively evaluates marital functioning at a lower level than does the father. Parental attitudes and coping styles differ in the two parents: the mother shows emotional/avoidance behavior, while the father is more task-oriented. The mother’s attitude towards the child is more negative, especially in terms of non-acceptance, rejection, denial of autonomy, and inconsistency. Both parents display an adequate level of care and concern.

**Conclusions:**

The parents of an RSTS child require particular support, which should encompass the entire family system. This support should be predicated on an analysis of the family’s available resources, as well as the barriers and difficulties that prevent them from fulfilling their roles.

**Key words:** coping styles, parental attitudes, caregivers

## INTRODUCTION

Rubinstein-Taybi Syndrome (RSTS) is a rare autosomal-dominant genetic disorder, occurring in 1 out of every 100,000 to 125,000 live births (Hennekam et al., 1990; Hennekam, 2006). RSTS presents as a new mutation in the child, with the parents showing no clinical signs; the probability of a successive child born with RSTS is estimated at 0.1%, while the risk of transmission of the mutated gene by an affected parent is as much as 50% (Imaizumi & Kuroki, 1991, cited by Cantani & Gaglesi, 1998; Hennekam, 2006; Stevens, 2009). There are several case studies in the literature documenting the transmission of RSTS from parent to child (e.g. Hennekam et al., 1989; Marion, Garcia & Karasik, 1993; Petrij et al., 2000). The etiology of RSTS is associated with chromosome anomalies; it can be caused by a mutation of the CREBBP gene (40-60% of cases, see Petrij et al., 2000; Coupry et al., 2002; Bartsch et al., 2005; Roelfsema et al., 2005; Bentivegna et al., 2006; Zimmermann et al., 2007), or by a mutation of the EP300 gene (2.6-3.2% of cases – see Roelfsema et al., 2005; Zimmermann et al., 2007; Bartholdi et al., 2007). Bartholdi et al. (2007) have observed that the phenotype differs to some extent, in the case of persons with the latter mutation, from the more familiar pattern seen in the case of the CREBBP mutation; specifically, the former do not present with the deformations of the extremities that have historically been regarded as pathognomonic for RSTS. Coupry et al. (2002) compared phenotype differences between RSTS patients with and without the CREBBP mutation, and observed that the majority of their subjects with this mutation showed the Gothic palate, while none of them had the hallux valgus typical for persons without the mutation.

Since the first systematic report (Rubinstein & Taybi, 1963) on the set of symptoms that later came to be called RSTS, research and analysis has described a constellation of clinical features for this syndrome. Cantani & Gaglesi (1998) analyzed 732 cases of RSTS, and produced estimates of the percentage of patients who display particular clinical features of the syndrome (see Table 1). Patients with RSTS present with characteristic facial features: highly arched eyebrows, down-slanting palpebral fissures, long eyelashes, hooked nose with low hanging septum, broad nasal bridge, highly arched palate and mild micrognathia. The characteristic facial expression is particularly noteworthy – a grimacing smile with the eyes almost closed. Talon cusps at the permanent incisors can serve as a confirmation of the diagnosis, since they occur almost exclusively in persons with RSTS (Hennekam & Van Doorne, 1990, cited by Hennekam, 2006). Anomalies in the structure of the extremities are typical, especially broad thumbs and broad big toes; the thumbs and big toes can be radially deviated, and there is often terminal broadening of the phalanges of the fingers, persistent fetal pads, and clinodactyly of the little finger (Simpson & Brissenden, 1973; Sener, 1999; Hennekam, 2006). Growth retardation with poor weight gain during infancy is common, but in later childhood this symptom can metamorphose into obesity (Stevens et al., 1990, cited by Hennekam, 2006). A number of congenital ocular

anomalies can co-occur with RSTS, the most common of which are nasolacrimal duct obstruction, abnormalities of the cornea and retina, ptosis of the eyelids, congenital or juvenile glaucoma, refractive errors and coloboma (Felgenhauer, 1973; Nelson & Tallbot, 1989; Genderen et al., 2000). Sometimes there are congenital heart defects (Okoroma & Izuora, 1987; Stevens & Bhakta, 1995; Bartsch et al., 1999), joint hypermobility, and skin anomalies, such as hirsutism, keloid formation (see De Silva, 2002), naevus flammeus on the forehead (see Centeno et al., 1999), as well as anomalies in the structure of the respiratory organs (see Magillo et al., 2005).

The specific anatomical anomalies found in persons with RSTS produce various health problems that are likewise characteristic: feeding problems in infancy,

Table 1. Typical clinical manifestations in 732 RTS cases (from: Cantani & Gagliesi, 1998, p. 84)

Category	Symptom	Percentage
Birth	Severe feeding problems in infancy	76.3%
	Neonatal distress or respiratory infections in infancy	75.6%
Older age	Stature under 50th percentile	92.7%
	Stature under 5th percentile	77.5%
	Bone age under 50th percentile	73.6%
	Obstipation	59.4%
Face	Head circumference under 50 <sup>th</sup> percentile	95%
	Narrow palate or palate high in appearance	93%
	Palpebral fissures with anti-mongoloid slant	90%
	Beaked nose	87.0%
	Microcephaly	87.0%
	Visible hypertelorism	83%
	Broad nasal bridge	81.8%
	Nasal septum alae	80.0%
	Grimacing smile	76%
	Ears abnormal in position, size, rotation, or shape	75.7%
	Deviated nasal septum	71.7%
	Micro/retrognathia	70.0%
	Heavy or highly arched eyebrows	69.6%
	Epicanthi	68%
	Dental anomalies	67.4%
	Strabismus	60.7%
	Small mouth	59.3%
Long eyelashes	57.8%	
Trunk	Stiff gait	83.8%
	Vertebral anomalies	75.3%
	Hypotonia, lax ligaments and hyperextensible joints	71.6%
	Pelvic anomalies	60.7%
Limbs	Sternal or rib anomalies	57.3%
	Thumbs and big toes with broad terminal phalanges	99.0%
	Other fingers with broad terminal phalanges	74.0%
Other anomalies	Fifth finger clinodactyly	50.3%
	Motor, mental, language and social retardation	98.5%
	Incomplete or delayed descent of testes	82.0%
	Hirsutism	76.0%
	I.Q. under 50	73.0%
	Cryptorchidism	67.0%
	EEG abnormalities	57.6%
Capillary hemangioma	56.6%	
Kidney anomalies or disease	50.7%	

respiratory problems, apnea, a tendency to dental caries, orthopedic problems, a tendency to constipation, difficulties under anesthesia (Bozkirli et al., 2000), and kidney problems (Hennekam, 2006). Persons with RSTS also incur an increased risk of tumors, especially meningioma and other brain tumors, as well as leukemia (see Felgenhauer, 1973; Miller & Rubinstein, 1995; Kurosawa et al., 2002; Kim et al., 2004).

In the cognitive domain, persons with RSTS are characterized by intellectual disability. Several studies have found an average IQ between 35 and 51 (see Hennekam, 2006; Stevens, 2009). Behaviorally, these patients present with short attention span, motor stereotypes, and poor coordination (Galera et al., 2009). Among the personality characteristics often found in RSTS are stubbornness, persistence, a strong sense of entitlement, and sudden mood changes, more pronounced with age. A very characteristic and doubtless favorable feature is sociability and ease in making social contacts, (Hennekam, 2006; Stevens, 2009).

Cytogenetic and molecular studies are needed in order to verify the diagnosis of RSTS, based on the discovery of pathognomonic dysmorphic features (Wallerstein et al., 1997; Stevens, 2009; Kim et al., 2010). As Hennekam states (2006), cytogenic and molecular anomalies occur in 55% of patients with RSTS, which means that the remaining 45% of cases, despite numerous genetic studies on this syndrome (see Hennekam et al., 1993; Petrij et al., 2000; Bartsch et al., 2002; Roelfsema et al., 2005; Bartsch et al., 2005), are diagnosed on the basis of clinical features (cf. Fig. 1). The more precise the information about the genetic basis, the more exactly the physical features of a given person can be anticipated, as well as the probability of various co-occurring disturbances.

This picture of the functioning of a person with RSTS, with numerous health problems, as well as behavioral and emotional disturbances and intellectual impairment, is consistent with the definition of a person with disability (Ossowski, 2008). The disability of one family member affects the functioning and QOL of the entire family system (Szarkowicz, 2009), and the parents are the persons responsible for the emotional climate prevailing in the family (Belsky, 1984, cited by Kersh et al., 2006). In recent years many researchers and clinicians have been studying the Health-Related Quality of Life (HRQOL) of persons with a disability, as well as their parents. The concept of quality of life has developed against the background of discussions surrounding the WHO definition of „handicap,” and has evolved over the last nearly half a century. Its application was initially limited to political and economic aspects of life, in the absence of illness. A biomedical model was then applied to health status assessment, and QOL came to be understood as an objective entity related to handicap: autonomy, work, income, and social activities. Over the last 20 – 30 years, the concept has moved from handicap to the individual’s subjective “well-being,” leading to the evaluation of one’s feelings about one’s own life (Tazopoulou et al., 2005; Błachnio, 2011)

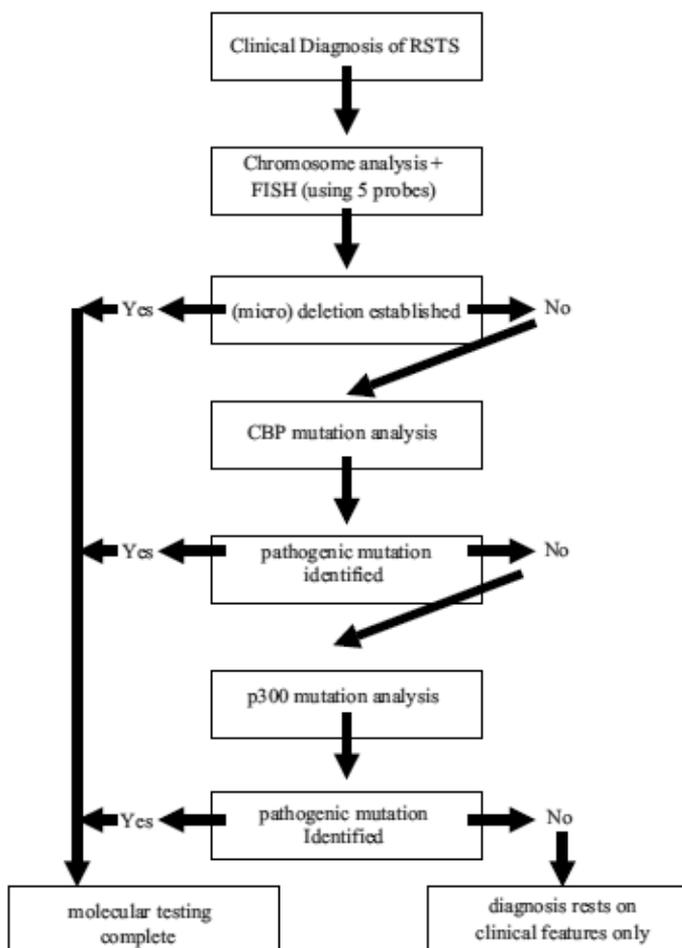


Fig. 1. Diagnostic strategy for RSTS. A microdeletion at #16p13.3 or a mutation in CBP/p300 can be found in about 55% of cases, leaving the diagnosis in 45% of the patients to rest on clinical features only. From: Hennekam, 2006, p. 981.

One reason for taking an interest in the QOL of the parents of children with disability, for reinforcing their parenting skills, and for promoting cooperation with specialists, is the principle of early intervention, with emphasis on the fact that care for a child with a developmental disorder gives the best results when it is provided in the family environment (Turnbull et al., 2000, Blue-Banning et al., 2004, cited by Wang et al., 2006).

QOL is a complex, multidimensional construct, and has been defined and conceived in many different ways in the literature (cf. Azouvi et al., 2005; Owczarek, 2010; Bidzan, 2011; Tomaszewski & Mańko, 2011). In our own work we refer to an individualized and subjective concept of QOL, which we understand as “the way in which each person evaluates their state of physical health, well-

being, self-sufficiency and lack of dependency, as well as the quality of their relations with the environment, and the state of their personal opinions and convictions” (WHO, cited by Kirenko, p. 18). The QOL of parents whose child has a disability, according to the conceptual model of QOL proposed by Schalock, Keith, and Hoffman, can be considered on three levels: intrapersonal, interpersonal, and social. The subjective evaluation of events on each of these levels, resulting from how they are perceived, conditions the mobilization of internal and external means of coping (Wyczesany, 2006).

The family in which a child is born with RSTS certainly becomes a “disabled” family, requiring special care (Urmańska, 2007), and numerous events affect the quality of life of its members. The parents go through a serious crisis, which is accompanied by a number of negative emotions, such as anger, guilt, bitterness, worry, and anxiety about the future (Twardowski, 2008; Sekułowicz, 2007). Disturbances in parental functioning often occur; they can manifest themselves in many domains and can assume many different forms. On the physical level these can include sleep disturbances, eating disorders, headache, or chest and stomach pains; on the psychological level there may be short-term memory loss, difficulties in concentrating attention, a feeling of disorientation, difficulties in making decisions, and obsession with the diagnosis; on the emotional level there can be a feeling of being overwhelmed by many different emotions and a feeling of loss of control over them, brief flashes of anger and dramatic changes in emotional reactions; on the spiritual level there is often a feeling of resentment to-

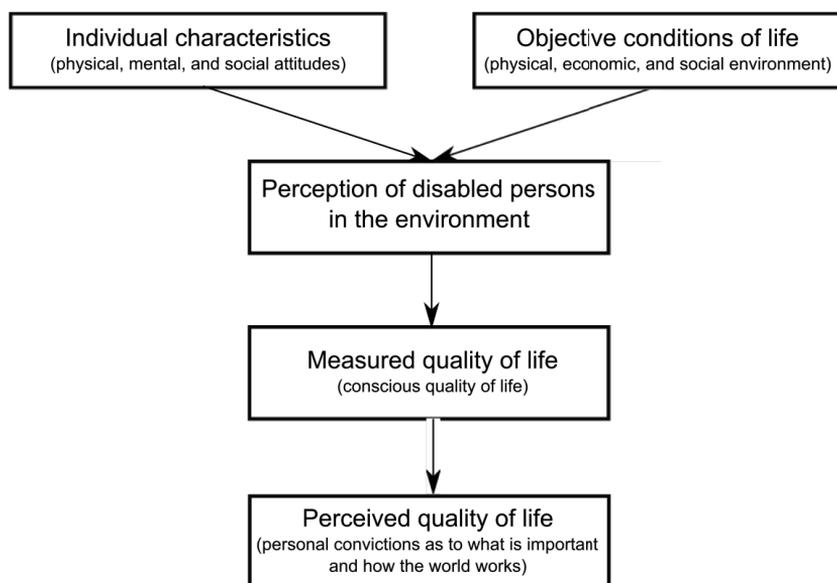


Fig. 2. The conceptual model of quality of life by R. Schalock, K. D. Keith, and K. Hoffman (cited by Wyczesany, 2006)

wards God, questioning of the reasons for the child's disease, and the loss or strengthening of faith (Friehe, Bloedow & Hesse, 2003, cited by Sekułowicz, 2007). A disabled child in the family is a major source of stress, which also affects the perceived quality of life. Researchers have emphasized that all parents experience stress caused by the arrival of the child, but the parents of the disabled child experience a greater number of stressful situations in daily life (Dumas et al., 1991; Lessenberry & Rehfeldt, 2004). The parents' emotional strength and their way of reacting to stress is essential in this context (Wyczesany, 2006). In order to adapt and to cope with this crisis situation, the parents are forced to make many changes. They must find themselves in a new situation, discover effective ways of coping with difficult situations, and learn to be the parents of a disabled child. This is a process. Twardowski (2008) describes the process of adapting to being the parents of a disabled child as consisting of:

- the shock phase;
- the emotional crisis;
- specious adaptation;
- constructive adaptation.

This last phase is characterized by an evolution from emotional collapse, grief, despair, anxiety, hopelessness, loneliness, and guilt, through the use of various defense mechanisms, such as denial, searching for the miracle cure, or looking for someone to blame, to acceptance of the situation and the child as they are, efforts to solve problems rationally and to provide optimal support for the child's development. As the author of this concept points out, the phases presented above constitute a model, and the situation faced by each family should be considered on an individual basis, with due regard for the fact that not every parent reaches the stage of constructive adaptation.

The functioning of a family with a disabled child, the level of stress felt by the parents, and the quality of life of the family are affected by many factors, some of which are associated with the child, some with the parents, and some with socioeconomic conditions. The most important factors affecting the functioning of such a family include:

- the child's age, which determines the types of tasks that the family must perform;
- the stage of the child's development at which the disability appeared;
- the type and severity of the disability;
- the specific nature of the child's functioning conditioned by the disability.

The extent to which the disability is visible is significant for the parents' quality of life and the amount of stress they experience, since society is dominated by attitudes unfriendly to disability, not accepting either the child or the parents. Another important aspect that determines the parents' experience is the way in which they find out that the child has a disability (Twardowski, 2008; Sekułowicz, 2007).

Among the factors associated with the situation of the parents of a disabled child, including a child with RSTS, are the following:

- the approach to life;
- the accepted value system;

- life goals;
- the place of residence (urban or rural), which affects access to specialized care and the degree of acceptance of the child's disability in the community.

The socioeconomic status of the family, which is affected by the type of work performed, education, and salary level, also has great significance for the functioning of the family. In the literature many reports can be found regarding the poorer material status of families with a disabled child (e.g. Kawczyńska-Butrym, 1966; Pisula, 1998; Lachewicz, 2006). The authors attribute this to the large financial expenditures required for rehabilitation and specialized care, as well as the change of vocational status of one of the parents, who gives up professional activity in order to devote full time to the child's care and rehabilitation. These financial difficulties are often a source of conflict and frustration for both the parents and the remaining members of the family, which further reduces the quality of life of these families (Twardowski, 2008; Sekułowicz, 2007).

When considering the quality of life of the parents of a disabled child, we should also pay attention to the factors resulting from the specific nature of the changes taking place in the family system at successive stages of family life. One such factor is family structure, defined by the number of family members, their age and gender, their social position in the family, etc. Such events as the arrival or departure of a family member, which are associated with a number of emotions, modify the family system and the evaluation of the quality of life. Another factor that affects the parents' evaluation of the quality of their lives is the performance of the specific functions assigned to the role they play in the family. The proper performance of these duties leads to the satisfaction of their own needs by the members of the system, which produces a feeling of security and satisfaction with belonging to a given family. The next factor is the degree and scope of involvement of the members of the family in the realization of roles and tasks in social systems outside the family, i.e. the performance of roles in the workplace or in social organizations. The skillful combination of roles and the performance of obligations resulting from various duties can be the source of pride and satisfaction, and thus of a high quality of life. In addition, family relations and communication constitute an important factor affecting the evaluation of quality of life. The optimal situation occurs when the family is held together by bonds based on love, goodwill, understanding, and acceptance, and the partner style of communication is dominant, which supports the expression of feelings, the revealing of personal experiences, and the display of respect and understanding for the communication partner (Rostowska, 2008).

Attention should also be given to environmental and contextual variables, and to the quality of life of persons nearby, who also affect the quality of life of the family (Dennis et al., 1993; Soresi et al., 2006; Turnbull et al., 2000, cited by Soresi et al., 2007; Tomaszewski & Mańko, 2011). An important modifier of the quality of life is social support, especially the perception of support provided by social and health services (Soresi et al., 2007).

The evaluation of the quality of life of parents with a disabled child when the disability is genetic in nature is important, since it provides information regarding the effectiveness and means of the support that parents receive and the choice of the optimal instruments for working with the family, so as to raise its quality of life, if necessary, and indirectly also to provide information on how to help the child, since the family is the first and the most basic place for the development and treatment of the child (see for example Lessenberry & Rehfeldt, 2004; Brown et al., 2006).

The goal of our work was to describe the specific nature of the functioning of the family of a child with RSTS in reference to the evaluation of the quality of life of the child's parents. Measures were chosen that are associated with the parents' quality of life:

- an evaluation of the child's psychomotor development and temperament;
- sociodemographic variables;
- psychological variables (such as parental attitudes and styles of coping with stress).

## **MATERIAL AND METHODS**

The following research methods were applied:

- the authors' own history questionnaire;
- participatory observation;
- analysis of medical documentation;
- the Self-Anchoring Ladder (Czapiński, 1992);
- the Life Satisfaction Scale (Juczyński, 2001);
- the Good Marriage Questionnaire, developed by M. Plopa and J. Rostowski (Plopa, 2008a,b);
- the Parental Attitudes Scale (Plopa, 2008a,b);
- The "Coping in Stressful Situations" Questionnaire (CISS), developed by N.S. Endler and J.D.A. Parker (Strelau et al., 2005);
- the EAS Temperament Questionnaire, developed by A.H. Buss and R. Plomin (Oniszczenko, 1997);
- the Bayley Scales of Infant Development, 3rd edition (Bayley, 2006).

## **CASE STUDY**

The family to be described here is a complete family, a traditional marriage, consisting of the parents (father, age 36, and mother, age 33) and three children, including two healthy children, 2 weeks and 2 years old, and a girl with RSTS, age 5. Both parents are university graduates and are active professionally. The family lives in the Tri-Cities region (Gdańsk, Sopot and Gdynia, on the Baltic coast of Poland). Their material status is good and their housing situation is satisfactory.

When examined, the girl with RSTS was 5 years old. At the age of 2;8 she had come under the care of the Psychological and Educational Clinic because

of delayed psychomotor development caused by RSTS. Since the age of 2;10 she had been participating in individual sessions with the psychologist, and was also included in the Early Support for Development program, where she had individual and group sessions with a speech therapist, a psychologist, a physical therapist, and a specialist in alternative and augmentative communication. Since the age of 3;5 she has been attending a special preschool on the basis of a diagnosis of Psychological and Educational Clinic with motor disability.

According to documentation from the genetics clinic, an analysis of chromosomes using the FISH method with RP11-461A8, RP11-75P12, and RP11-95J11 probes for the region critical for RSTS, i.e. 16p13.3, did not reveal a deletion in the tested region.

An x-ray examination of the patient's feet and hands at the age of 1;4 did not show any obvious pathologies of the bones on either side, as far as the shape and dimensions of the calcified portions of the bones of the fingers, mid-hand, and wrists were concerned. The skeletal age corresponded to her calendar age. There was an additional bone in the left foot between the fourth and fifth bones of the mid-foot. The shape and size of the remaining bones were normal. The clinical features, which included delayed psychomotor development, heart defects, rectal defects, polydactyly, thickening of the thumbs and big toes, and dysmorphism justify a diagnosis of RSTS. The absence of a deletion in the critical region does not preclude such a diagnosis, since these deletions have been identified in only about 10% of patients with RSTS. At the present time there is no possibility of performing other tests in Poland that would confirm the clinical diagnosis.

On the basis of individual therapy with the patient, two meetings a week over a period of three years, she has been observed to have delayed psychomotor development. A detailed functional analysis has been presented in Table 2.

The patient is sociable, mild-mannered, and makes contact easily with both adults and children. In her preschool group she initiates and coordinates thematic play with her peers. She is likable, enjoys being in the center of attention, and often attracts the attention of adults by her behavior. Emotional lability is characteristic. She is internally motivated, but due to her short attention span she requires reinforcement and support from an adult in order to complete an activity. Among her undesirable behaviors are selfishness and beating her peers.

The physical features typical for RSTS include stiff hair, eyelid slits pointed downwards, a narrow jaw (micrognathia), small head, gothic palate, a characteristic smile with grimace and closed eyes, a hooked nose with the septum extending beyond the nostril, a broad nose base, ears placed lower than normal, thick eyebrows, a small oral cavity, thumbs and index fingers with broad fingertips, and clinodactyly of the little finger.

The patient has frequent upper respiratory infections, heart disease, disorders of visual refraction (she wears glasses with a strength of +2.5), and little or no control of vegetative functions associated with eating.

Table 2. Description of the child's functioning, based on the Bayley Scales of Child Development – Third Edition

Sphere	Functioning
Gross motor	moves independently, can go up and down stairs with support, placing her feet on one step; stiff gait on wide stance; walks backward; runs slowly with poor coordination; can bend over and pick up a toy; throws a ball inaccurately, kicks the ball; stands on right and left leg with support for several seconds (age 21 months)
Fine motor	due to the specific build of the hand the pincer grip is imprecise; drawings are scribbles; imitates the drawing of vertical lines, horizontal lines, and circles, using her hand to hold the page in one place; more often uses the right hand; builds towers of 10 blocks, builds a train; joins and separates constructions of blocks; imitates hand movements; differentiates by touch the most important shapes without visual control (age 34 months)
Communication	<p>Receptive communication:                      Recognizes the names of clothing items, body parts, basic objects, animals, activities; understands the use of objects from daily living; understands the basic personal pronouns, and the prepositions "through, around, on"; understands size markings; understands nouns in different grammatical cases; understands the concepts of "one" and "more" (age 35 months)</p> <p>Expressive communication:                      uses various combinations of 2-3 words; names objects familiar from daily living and several activities (e.g. "reads, sleeps, eats") , often supported by a Makaton gesture; in familiar situations she can answer a simple question coherently; asks "What is that?"; uses the pronouns "I" and "you" (age 30 months)                      uses alternative and supportive means of communication: a communication book with PCS symbols, Makaton gestures</p>
Cognitive functions	assembles a picture composed of two elements; matches similar pictures; completes a puzzle putting all the elements in the right place within the allotted time; understand the concept of unity; groups and matches objects according to dimensions; recognizes objects in incomplete pictures; play developed to the level of make-believe (age 32 months)
Independence	Eats independently using a spoon and fork; drinks from a cup independently, with occasional spills; wears diapers, does not report physiological needs. Toilet training in progress for a year.
Social development	Mild-mannered child, tries to make contact with other people; cooperates with adults; displays joy at the sight of people she is close to; displays shyness in the presence of strangers; sensitive to verbal praise and censure.

An analysis of the child's temperament using the EAS Temperament Questionnaire by A.H. Buss and R. Plomin indicated:

- high scores on the emotionality scale (80th percentile), which indicates a tendency to quick and intensive reactions of uneasiness, difficulty in maintaining calm, and high sensitivity to stressful stimuli;
- medium scores (50th percentile) on the activity scale, pointing to average manifestation of motor activities, as well as average strength and intensity of reaction;
- medium scores (60th percentile) on the sociability scale, which indicates average intensity of looking for contact with other people and remaining in social situations;
- low scores (30th percentile) on the shyness scale, suggesting a lack of anxiety in the presence of strangers.

Table. 3. Parents' subjective perception of the functioning of their daughter in the present and the future

Parents' perception of the functioning of the child	Present	Future
Significantly deviates from the norm		
Slightly deviates from the norm	X	X
Functions inormally (like peers)		

An analysis of the interview questionnaire provided information regarding both parents' subjective perception of the functioning of their daughter in the present and the future (see Table 3).

Both parents evaluate their daughter's functioning in both temporal horizons as only slightly deviating from the norm.

For the purposes of the present study, it was important to evaluate the quality of life and satisfaction with life of the parents of this RSTS patient. We used two methods: the Self-Anchoring Ladder (to evaluate quality of life) and the Satisfaction with Life Scale (SWLS). The results of the SWLS are closely related to the evaluation of the quality of life, which is why many researchers use it to make conclusions not only regarding the level of satisfaction with life, but also regarding quality of life.

An analysis of the results from the Cantrill Ladder indicates that both parents evaluate the quality of their life as average in the present temporal horizon. Taking into account the quality of life in the best and worst weeks of their life, it can be seen that this family, like every average family, has good and bad moments, and evaluates them adequately in terms of the intensity and valence of the emotions. In addition, both the mother and the father are optimistic in respect to their future, evaluating it one point higher than the present. Attention should be drawn to their divergent assessments of the past. When evaluating her life five years ago, the mother assigned it a high value in comparison to the present. This evaluation coincides with the birth of their first child, who is disabled. The father, on the other hand, currently evaluates the past as average in terms of satisfaction, like the present. The detailed results obtained from this test are given in Fig. 3.

The results obtained from the SWLS questionnaire, which are presented in Table 4, provide further information regarding the subjective evaluation of the quality of life by these parents. In response to the statement, "If I could live my life over again, I wouldn't want to change hardly anything," both the mother and the father stated that they did not agree with this statement, indicating that they would make changes in their life. Some of their responses to the statements were elusive. On a scale from 1 ("I completely agree") to 7 ("I completely disagree") they chose 4 ("I neither agree nor disagree"). To the statement, "I am satisfied with my life," both parents replied in the affirmative. In the global score

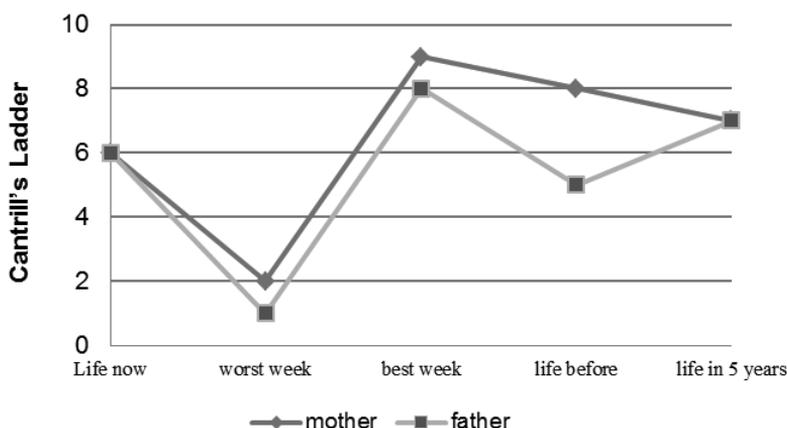


Fig. 3. Results obtained from Cantrill's Ladder by the parents of a child with RSTS, on a scale of 0-10, where 0 is "the worst life I could expect" and 10 is "the best life I could expect"

Table 4. Results obtained from the SWLS by the parents of a child with RSTS

Statement	Mother	Father
In many respects my life is nearly ideal.	I disagree	I neither agree nor disagree
The conditions of my life are perfect.	I disagree	I neither agree nor disagree
I'm content with my life.	I agree	I agree
I've achieved the most important things I've wanted in my life	I neither agree nor disagree	I agree
If I could live my life over again, I wouldn't change hardly anything.	I strongly disagree	I agree

from the SWLS the mother had a low score for satisfaction with life (40<sup>th</sup> percentile), whereas the father had an average score (60<sup>th</sup> percentile).

The evaluation of the marriage affects the evaluation of the quality of life. We evaluated this sphere by using the Good Marriage Questionnaire. The results are shown in Fig. 4.

The Good Marriage Questionnaire covers four dimensions: intimacy, disappointment, self-realization, and similarity. The scores obtained on the intimacy scale by both parents indicate an average degree of intimacy in their perception. On the disappointment scale, the mother's result indicated that she feels a high level of disappointment with the marriage, while the father's score reflected an average feeling of disappointment with the marriage and autonomy. As for the self-realization scale, the mother is convinced that she cannot realize herself in her marriage, while her partner has an average feeling of the possibility of self realization. Both parents similarly perceive an average feeling of similarity in regard to the realization of marital and family goals.

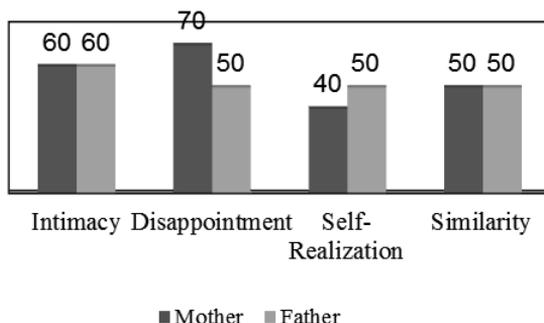


Fig. 4. Percentile results from the Good Marriage Questionnaire obtained by the parents of a child with RSTS

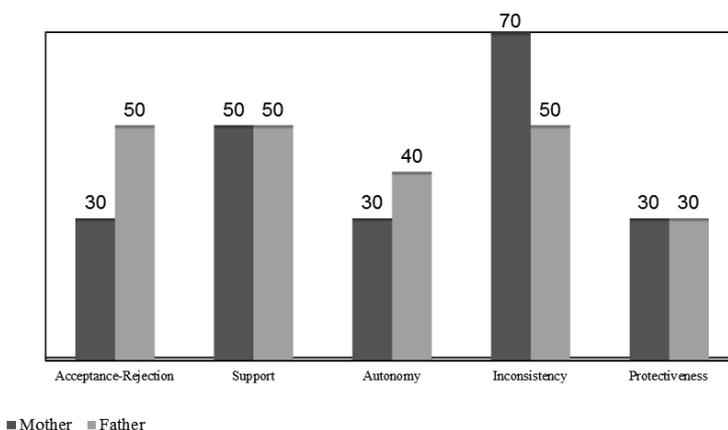


Fig. 5. Percentile results from the Parenting Styles Questionnaire obtained by the parents of a child with RSTS

On the Acceptance-Rejection scale of parental attitudes test, the parents showed differing attitudes. The mother’s score tended to be lower, indicating a lack of acceptance of the child, and also some rejection. The father’s score, on the other hand, was in the normal range for acceptance. On the Requirement scale, both parents displayed a reasonably correct attitude. On the Autonomy scale, both parents had low scores, indicating a lack of autonomy in relation to the child, while on the Consistency scale the parents displayed diverging attitudes. The mother’s score indicated a lack of stability, emotional lability, and inconsistency in relation to the child (an incorrect attitude). The father displayed a reasonably correct attitude on the Consistency scale. An analysis of the Protectiveness scale indicates that both parents apply this parenting style. The low percentile scores they achieved on this scale point to a correct attitude, characterized by adequate care and concern for the child. The percentile scores for the parental attitudes displayed by the mother and father are shown in Fig. 5.

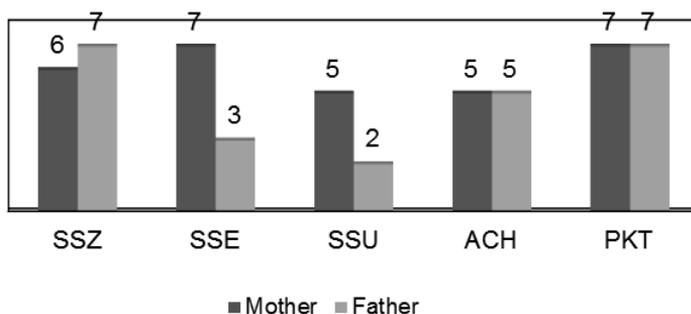


Fig. 6. Percentile results from the CISS Questionnaire obtained by the parents of a child with RSTS. Legend: SSZ = task-oriented; SSE = emotional; SSU = avoidance; ACH = substitute activities; PKT = social contacts

The birth of a child with disability is associated with a high level of stress and psychological burden, which affects the evaluation of quality of life by the child’s caregivers, most often the parents. Dealing with the difficulties that result from being the parent of a disabled child activates different styles and strategies for coping in different people. Thus it was important for us to analyze the styles of coping with stress displayed by the parents of this child with RSTS. Figure 6 presents a comparison of the results for both parents in respect to coping styles.

On the test of ways of coping with stress the two parents displayed differing results. The mother most often uses two styles (concentrated on emotions and avoidance), while the father shows a task-oriented style.

## DISCUSSION

The quality of life of this family of a child with RSTS does not differ from that of other families with healthy children. Both parents evaluate the quality of their lives as average, and this family, like every other, has good moments and bad ones. The results we obtained are inconsistent with a number of reports from research on families having children with a disability, claiming that they have a marked lower quality of life, or other parameters indirectly associated with it, such as mental health (Harris & McHale, 1989; Blacher et al., 1997, cited by Kersh et al., 2006), level of stress (Beckman, 1991, Cameron et al., 1991; Dyson, 1997, Baker et al., 2003, cited by Kersh et al., 2006; Forde et al., 2004) or satisfaction with marriage (Friedrich & Friedrich, 1981, cited by Kersh et al., 2006; Kersh et al., 2006), in comparison to families that have healthy children.

In the literature one can also find research whose results, like our own, show no differences in the subjective evaluation of the quality of life or satisfaction with life. The parents of disabled children examined by Wang et al. (2006) evaluated the quality of their life relatively high. Some researchers have also found no differences between mothers and fathers in this respect. The results of other studies (Bristol et al., 1988; Hoare et al., 1998) indicate no essential differences in

the symptoms of depression among the mothers of disabled and healthy children. Turnbull et al (1993, cited by Kersh et al., 2006) and Kersh et al. (2006) in their analysis did not observe any differences in the level of parental stress or manifest symptoms of depression among mothers and fathers of disabled children in comparison to the general population, while Donovan (1988, cited by Kersh et al., 2006) reported no differences in respect to marital satisfaction between families with disabled children and those with healthy children. Yet other research reports (Soneman & Gavidia-Payne, 2006, cited by Kersh, 2006) suggest that the mothers and fathers of children with various kinds of disability experience from average to above average satisfaction from their marriages, and other researchers have reported no association between the severity of the child's disability and marital adaptation.

One possible explanation of the average quality of life score in comparison to the general population for these parents of a child with RSTS may be their family resources.

When a child appears, the change in the family that occurs in respect to its structure, function, material situation, and social status significantly modifies the parents' quality of life, since it presents them with a number of challenges which are undoubtedly stressful (Brown et al., 2006). It often happens that the families of disabled children undergo a breakdown; this does not happen, however, to all families: the family we have described was complete. This family belongs to those which, despite difficulties, manage to overcome the challenges associated with raising and carrying for a child with a disability, to adapt to the situation, and some parents even manage to appreciate and find positive aspects of being the parents of a disabled child (Ferguson, 2002; Ylven, Bjorck-Akesson & Granlund, 2006; Scorgie et al., 1996, cited by Wilgosh & Scorgie, 2006). How a family copes with solving problems depends, among other things, on the resources at its disposal, that is, "the properties, attributes, and personal skills of the persons who are available to the family and can be used in coping with stressful situations" (Plopa, 2008a, p.41). This author states that it is possible, thanks to such resources, to reinforce the continuity and stability of the family, to overcome threats, and to adapt to changed circumstances. Resources are characteristic for each family; they are shaped under the influence of the family's experience, and can be divided into three main sources:

- resources associated with a particular member of the family, i.e. intelligence, consciousness, readiness to take up challenges, knowledge, skills, personality characteristics (such as responsibility and sense of humor), a feeling of control over one's own life, a feeling of one's own value, empathy, and state of health;
- resources consisting in the family as a whole, such as harmony, the clarity and consistency of the roles and principles that are dominant in the family, the internal coherence of the family (i.e. trust, support, and mutual appreciation), integration and respect for individuality, adaptability, coping strategies, communication skills, the family's strength (i.e. a feeling of control over events,

an active approach to any difficult situations, and the faith that change will have a positive effect), the amount and form of time the family spends together (which indicates its integration and stability), the family's ability to use individual resources for coping with stress and to solve problems, and the material assets the family can use;

- resources associated with social support provided by the community in which the family lives, including individuals, groups, institutions, and associations (Szarkowicz, 2009).

Research on the families of children with developmental disorders has confirmed these assumptions about family resources, i.e. the assets that protect them from crises. On the basis of numerous observations (Scorgie et al., 1996, Scorgie et al., 1997, Nota et al., 2003, Wilgosh et al., 2004, cited by Wilgosh & Scorgie, 2006), the picture has emerged of the parents of a disabled child who effectively cope with difficulties ("effective life managers"), and are distinguished by the following characteristics:

- positive life management strategies, such as positive reframing of thinking and circumstances, and maintaining equilibrium between the obligations of work and family;
- personality features, such as constructive decision making, problem-solving skills, and strong personal convictions;
- strong positive personal and social results of transformation, i.e. revaluing of what is important in life (Wilgosh & Scorgie, 2006).

Alston & McCowan (1994, cited by Wilgosh & Scorgie, 2006) & Alston & Turner (1994, cited by Wilgosh & Scorgie, 2006), as a result of research conducted in a population of African Americans, created a model of the assets of a family, in which they distinguished four key elements:

- strong faith/religious affiliation;
- kinship bonds;
- elasticity in the performance of roles;
- emphasis on education and the work ethic.

Research indicates that family resources have a positive effect on adaptation to major health problems, thanks to the skills of coping and adapting (Wallander & Varni, 1998; Downe-Wamboldt et al., 2006, Rolland & Walsh, 2006, cited by Hsieh et al., 2008). Hanline & Daley (1992, cited by Wilgosh & Scorgie, 2006) observed that the internal coping strategies of the family are better predictors of the family's strength than the use of social support.

The family described here is characterized by a number of features that can be qualified as family assets. In respect to the resources of single family members, both the mother and the father exhibit significant intellectual potential, as indicated by their college education. The parents, especially the mother, have acquired broad knowledge regarding the RSTS syndrome and are aware of the limitations that result from it. Both parents display moderate optimism in regard to their future, which may indicate a realistic evaluation of their situation. Among the resources of the family as a whole, we should include the fact that the family

is a complete, traditional marriage, with two healthy children, and both parents are professionally active, which means that they can find fulfillment in a role other than parenting to develop themselves and to change their routine of daily life.

Another resource is the family's place of residence: a large city, which provides greater access to specialists, as well as greater possibilities for their daughter's rehabilitation. They also have satisfactory housing. In the case of a person with disability, economic factors are essential. These parents state that the family's material status is good.

Both parents perceive an average feeling of similarity in respect to the realization of marital and family goals, and their ways of coping with stress are complementary: the father usually employs a task-oriented style, while the mother is more emotional. As for social support, the family receive support from the extended family (especially the grandparents), as well as institutions (their daughter attends a public special preschool), which undoubtedly has an effect on the parents' higher evaluation of the quality of life.

It should also be emphasized that despite the child's delayed development, characteristic appearance, and motor disability, the parents report that the reactions of people in the community to their child are the same as their reactions to their healthy children. The girl herself does not display any exceptionally difficult behaviors, and her typical characteristics (such as sociability, good nature and facility in making contact with adults and children) cause her to be kindly received by the people she meets.

The divergencies we discovered between the scores of the mother and the father in respect to satisfaction with life and marital contentment are supported by numerous comparative studies of the fathers and mothers of children with developmental disabilities. Much of this research (Bristol et al., 1988; Veisson, 1999, cited by Kersh, 2006; Olsson & Hwang, 2001; Trute, 1995, cited by Wang et al., 2006) indicates that, in comparison to fathers, the mothers of disabled children present more symptoms of depression. The results of research by Friedrich & Friedrich (1981, cited by Kersh et al., 2006) suggest that the mothers of children with developmental disabilities evaluate their marriages as less satisfying than the mothers of healthy children, while Wallander & Varni (1998) found that these mothers have lower mood, problems with adaptation, and more problems with mental health. These researchers also observed, however, that despite these difficulties, hope and social support can have a positive effect on the adaptation of the mothers of children with developmental disabilities.

The father described here, in all the domains we studied - marital contentment, quality of life, and satisfaction with life – achieved average scores in comparison to the general population. The mother, on the other hand, evaluated her satisfaction with life rather low; she feels a high level of disappointment with the marriage, and in respect to self-realization, she is convinced that she cannot realize herself in the marriage. Taking into account the mother's higher evaluation of her life 5 years ago, which coincides with the birth of her first child (who is disabled), along with her possible desire to make changes in her life and her lack

of acceptance of her child, it can be assumed that the lower general satisfaction with life is associated with motherhood in general, or being the mother of a disabled child in particular.

One of the ways of improving satisfaction with life can be social support. As indicated by research (Jokinen & Brown, 2005, Seltzer et al., 2004, Soresi et al., 2006, cited by Soresi et al., 2007), in order to reduce the difficulties of parenting a disabled child, families with a disabled child should be provided with various kinds of support and skill training throughout the child's life. In addition, this should be support that reduces the negative effects of disability and reinforces the positive (Summers et al., 2005, cited by Soresi et al., 2007). Soresi et al., 2007 analyzed the effectiveness of an intervention consisting in a cycle of training courses for parents, the goal of which was to emphasize autonomy and reinforce coping skills, problem-solving skills, and assertiveness, in order to enable the family to take full advantage of the resources they possess. The results obtained by these authors indicate that the knowledge acquired by the parents persisted for a long time and was generalized; the parents showed a higher evaluation of their satisfaction with family relations and the support they were receiving in comparison to the situation before their training. The intervention also had a positive impact on the perceived quality of life of the family, the use of knowledge, and the well-being and contentment of the participants. The undoubted advantages that result from early detection and early intervention for children with developmental disabilities should also be mentioned, since these lead to improvement of the quality of life and reduce the negative impact of the child's disability on the family, both for the child and the caregivers (Hsieh et al., 2008).

## **CONCLUSIONS**

Our evaluation of the quality of life of the parents of a child with RSTS and the factors that condition it justifies the following conclusions:

1. Being the parent of a disabled child with RSTS has an effect on the evaluation of the quality of life in the past, the present, and the future, and on the evaluation of satisfaction with life.
2. The mother of this child with RSTS had a lower evaluation of marital functioning than did the father in all domains, that is, intimacy, disappointment with the marriage, self-realization, and similarity, which may be affected by their daughter's disability.
3. The parents of the child described here display differing parental attitudes. The mother shows more negative attitudes towards the disabled daughter than does the father, which involves especially lack of acceptance, rejection, lack of autonomy, and inconsistency.
4. Both parents show adequate care and concern for their disabled daughter.
5. These parents activate differing coping styles in difficult situations, including those associated with their child's disability. The mother most often uses an emotional style or an avoidance style, while the father is usually task-oriented.

6. The evaluation of the quality of life by the parents at an average level may be affected by the family's numerous resources (originating from the child, the parents, and the community).

## **PRACTICAL IMPLICATIONS**

The analysis of the quality of life of the parents of a child with RSTS, taking into account the functioning of both the disabled child and the parents, provides a number of practical implications. The most important of these are the following:

1. Families that have a child with a rare genetic syndrome require particular support, which results from the limited amount of information, specialists, and social awareness, due to the rarity of the syndrome and the still inadequate amount of knowledge and research regarding a significant number of genetic disorders.
2. Therapists with many different specialties should continually expand their knowledge in the subject of rare genetic disorders, their etiology, pathomechanism, and forms of rehabilitation.
3. Support and specialized care should be provided for the entire family system, not only the parents, but also the other children in the family and the grandparents, due to the fact that the quality of life of particular members of the family is mutually interdependent, as well as the fact that the family is the basic place for the child's development and therapy.
3. In the process of supporting a family with a child that is developmentally disabled, it is necessary to make a thorough analysis of the family's resources, knowledge, possibilities, and skills, as well as the barriers and obstacles that impede their performance of their tasks as parents.

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