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Support for the family and child with multiple congenital anomalies in the contemporary reality

Issues concerning the functioning of a person with a multiple disability, complex syndromes of organic anomalies (often of a congenital and genetic nature) have acquired, especially in recent years, an exceptional dimension as well as theoretical and practical significance. This is a consequence of the fact that modern special education is to a much greater extent interested in the subject so far discussed rarely or never. Another reason is a noticeable openness of contemporary society to dilemmas of people who require specialist assistance and an interdisciplinary support, especially people who suffer from multirange limitations. Thus the subject of support for the family and a person with a multiple disability, with congenital anomalies, should be perceived not only within medical or psychopedagogic categories, but also humanistic and moral ones.

Medical definitions formulate the notion of a congenital developmental anomaly as "any external or internal morphological deviation from a normal development, irrespective of its pathogenesis, the time when it initially appeared and the time of diagnosis"[1]; and from a medical point of view the issues of congenital defects fall within the scope of dysmorphology.

From a pathomorphological perspective, congenital developmental anomalies are divided into:

1. Malformations – stemming from the influence of internal factors on the developmental process (e.g., cleft palate).
2. Disruptions – connected with the impact of external factors on the primarily normal developmental process (e.g., amniotic band syndrome, ABS).
3. Deformations – the outcomes of mechanical factors and their impact on the developmental process (e.g., club foot).
4. Dysplasia – the result of abnormal cell differentiation within a tissue (e.g., ectodermal dysplasia, ED)[2].

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[1] M. Wiśniewska, *Wrodzone wady rozwojowe*, www.kgm.amp.edu.pl, 22.07.2007.
[2] Ibidem

Another division of multiple developmental anomalies include the following categories:

1. Sequences – resulting from a single developmental defect or mechanical factor, e.g., Pierre-Robin Sequence (the primary defect is micrognathia).
2. Complexes – the effects of a disrupted development of a developmental area or part, e.g., abnormal positioning of the embryo's artery may lead to hypoplasia of bones and muscles within the reach of this artery.
3. Syndromes – multiple congenital developmental anomalies linked by a common pathogenesis (e.g., Edwards Syndrome, Gregg's Syndrome).
4. Associations – non-random complexes of anomalies whose components appear together more often than it would be randomly possible, e.g., VACTERL:

V – *vertebral anomalies*;

A – *anal atresia*;

C- *cardiac anomalies*;

TE – *tracheoesophageal fistula*;

R – *renal/urinary anomalies*;

L – *limb defects*[3].

The epidemiological division provides the following groups of congenital anomalies:

1. Multiple and isolated anomalies.
2. Major anomalies (disrupting the activity of the organism) and minor anomalies (without serious consequences).
3. Lethal anomalies and anomalies which do not impact the mortality.
4. Anomalies which appear in a given family and anomalies which appear sporadically[4].

Congenital developmental anomalies are caused by genetic factors (chromosome aberrations, mutations in single genes, multiple genes determinants), environmental agents (teratogens) as well as mixed ones. Etiology of 60% of serious congenital anomalies is unknown, whereas in 85% of congenital anomalies of known etiology the most significant role is played by genetic factors.

In many countries worldwide, including Poland, birth registrations of children with multiple congenital anomalies have been recorded for years now. However, the obtained data displayed are far from the actual state of affairs, although as early as 1972 the World Health Organization (WHO) recommended devising registers of genetic diseases, and in 1974

[3] Ibidem
[4] Ibidem

appeared the idea of creating a register of congenital anomalies concerning the territory of the European Union countries. In 1979, a special system called EUROCAT was formed, and on 01.04.1997 the Polish Register of Congenital Developmental Anomalies (Polski Rejestr Wrodzonych Wad Rozwojowych; PRWWR) was initiated, which became a part of the EUROCAT system on 01.06.2001.[5]

2. Problems of the family and of the child with multiple congenital anomalies

Due to the fact that multiple congenital anomalies are connected with the co-appearance of at least a few organic and developmental deficits, this condition consequently leads to complex and multidimensional functional effects. Thus the problems experienced by the child and family are also similarly complex and multidimensional. The question is which problems must be faced by the family and which ones by the child.

2.1. Family problems

A detailed and objective analysis of possible problems experienced by the family of a child with multiple congenital anomalies allows one to place these difficulties in several basic categories: 1) receiving information of the child's health condition and disability; 2) accepting these facts; 3) taking action to inform the closest relatives and friends about the situation; 4) taking action towards diagnosing the child's developmental disorders, improving his/ her health condition and his/ her rehabilitation; 5) accepting the fact of being a family with a child with a multiple disability as well as all the consequences stemming from it for the entire family as a whole and each particular member in it.

Having participated for many years in various types of rehabilitation courses and holidays organized for visually impaired children or children with a simultaneous visual and hearing impairment, as well as working as a special needs teacher and educationalist, I have had ample opportunities to talk to parents, especially mothers, about problems which they experienced during the first few years of their children's lives, including the prenatal, perinatal and postnatal periods.

On the basis of those conversations it is possible to identify certain circumstances of the prenatal period common in most cases, firstly the fact that examinations conducted by a specialist who supervises the pregnancy, e.g., USG, do not always allow the diagnosis of the presence of multiple anomalies; secondly the fact that even if developmental defects are

[5] Ibidem

noticeable, this information is not necessarily always passed on to the mother-to-be. This situation stems from a few reasons: lack of certainty as to whether an anomaly (or anomalies) is actually present; a physician's fear of a wrong diagnosis as well as of a mother's reaction on receiving information that the presence of anomalies is suspected in her child. Another reason is a physician's wrong attitude concerning their professional tasks, understood as formal control over the pregnancy and formally supervising it to its conclusion without considering the quality of the process. There are also imperfections in the sphere of obtaining from pregnant women information concerning the presence of disability in other children in the family, parents themselves or other, distant relatives.

A similar situation is observed in the perinatal period. Serious doubts, also those of an ethical nature, are raised by the way parents are informed about their children's condition and probable future. The process of providing information is too often superficial and chaotic. The seriousness of the situation is underestimated, whereas parents' feelings and emotions are not taken into consideration. As for the mother, the fact that her organism is weakened after the childbirth is overlooked. In addition, in the case of the presence of complex, multiple anomalies in a child, information on his/ her future life, and even survival, is provided, to put it mildly, without gentleness, straightforwardly, bluntly, and in a form that is almost like a decisive, fatal judgment.

Next months, and often on average the first two years, of such a child's life are connected with diagnosis, treatment and fighting for survival. For parents, this is a period of sleepless nights, nightmares, waking up suddenly from sleep. Moreover, this is a period of numerous and long-lasting hospitalisations in specialised institutions, often situated far from one's place of residence, which always results in the separation of child from family and often of both child and mother accompanying her baby. This is a period of stress resulting from diagnostic procedures and medical treatment, including surgical interventions, undertaken by physicians. This is a time of enormous physical effort, fear for the child's life, for what is happening back home, for the way the family and older children, left under the care of the father or grandparents, are coping. This is also a period of numerous phone calls, significant financial expenses, extraordinary emotions and anxiety for the child's well-being and for the present day. What tomorrow and the near future will bring seems somehow less important, and in many situations is beyond the parents' consciousness, is simply not on their minds. Thoughts about rehabilitation or specialist psychopedagogical therapy are not yet crystallized and sometimes do not even appear. In fact, there is no time for thinking about accepting the child's disability. Everything is dominated by activities of a medical nature, such as obtaining

medications or life-supporting equipment that is difficult to procure. The main issue is the child's survival. Only later, when the child's health condition improves, is there more time to pay attention to various thoughts flashing through one's mind. Now appear more or less clear visions of the child's rehabilitation and future state and concrete actions are undertaken to achieve the best results in improving the child's quality of life. Now specialist out-patient clinics which offer psychological, pedagogical or logopedic therapy are being sought. A new rehabilitation stage begins, i.e., psychopedagogic rehabilitation. Similar to the previous stage, i.e., medical therapy, this period is linked with physical effort, financial expenses, the continuation of medical treatment, lack of time, life on the run, keeping constant track of time in order not to be late for a particular appointment at a particular place. Additionally, there are domestic duties connected with managing the house, looking after the family and other children, which are difficult to combine with attending rehabilitation courses or going to specialist consultations which often take place on the other end of Poland. Consequently, a family atmosphere, mutual understanding, assistance and tolerance are of the utmost importance. And later there come decisions concerning pre-school education, a probable delay in school education, as well as choosing a specific educational institution. Again there appears an internal dilemma, the struggle with everyday problems, with thoughts whether and how the child will cope in a new situation. It also results in stress, frayed nerves, sleepless nights due to nightmares connected with these issues. This is, generally speaking, the picture of the first period of life in a family with a child with multiple congenital anomalies – constantly on the run, in stress, struggling with fatigue and thoughts of 'what next'? What, then, are the problems of such a child in the discussed period, since it seems quite clear as to the reasons.

2.2. Child's problems

When analysing the problems of a child with multiple congenital anomalies several significant issues need to be addressed. These include, among others, questions of a physical-physiological nature, i.e., communication with the nearest environment as far as fulfilling the primary and secondary needs is concerned, and then communication on a wider, social perspective (distant family, neighbours, and especially children of the same age group), as well as getting to know the reality and acquiring important coping abilities, skills and habits.

Physical-physiological issues are connected with activities performed by others for a child in order to improve a child's health condition (diagnostic examinations, pharmacological treatment, sometimes surgical intervention, medical rehabilitation). Such

situations undoubtedly involve significant physical effort, fatigue, physical pain, a necessity to undergo a variety of medical procedures which are sometimes quite painful.

Issues connected with fulfilling one's needs are linked with the dilemmas of communication within a close environment. Experiencing problems with acquiring traditional methods of communication, a child with congenital defects is not always able to send messages clearly and understandably. This leads to disruptions of communicative, interpersonal and psycho-emotional relations between the child and the close family members with respect to fulfilling the child's needs.

The lack of, or serious limitations, in using articulated speech and non-verbal communication results in complications in the sphere of relations with a wider social environment. This concerns primarily members of a distant family, neighbours, and most importantly, peers. On the one hand, the child needs and strives for contacts with these people (which is quite natural); on the other – the child has problems, which stem from developmental defects, in beginning, developing, retaining and modifying – depending on the circumstances – proper and mutually understandable communicative interactions and even psycho-emotional ones. The child struggles between comprehending and not being able to comprehend personal intentions and those of others. The child experiences numerous failures when movements, gestures and produced sounds are not properly understood by others. The most serious failure is, however, the inability to acquire them in such a way that they might be interpreted accurately. This situation becomes a source of many experiences involving traumatic effects, conflicts, lack of success, which can all contribute to isolation and the shutting of oneself away from others.

Another type of dilemma involves issues concerning the possibilities, and to be more specific, limited possibilities to understand the surrounding reality and to acquire important abilities, skills and habits. Children learn to know the surrounding environment by means of action and imitation, and in the analysed circumstances these means are limited or non-existent. Thus, only applying special methods of working with such a child, multiple repetitions of a given situation, presenting incentives which draw attention and organising the learning process by others provide some opportunity for such a child to realize what is happening in the surrounding environment, what the sense and meaning of it all is, and what needs to be done, i.e., what skills must be learnt, in order to be relatively self-dependent and to find fulfilment. This is only a sketchy description of important and significant problems that children with multiple congenital anomalies as well as their immediate families must cope with. Thus it is worthwhile to illustrate the dilemmas described above by means of a

specific, individual case of a child with congenital anomalies and her family. This will be done on the basis of Malwina K. – being a representative of a group of children with multiple disabilities.

2.3. *Problems of the family and of the child with multiple congenital anomalies – case study of Malwina K.*

Malwina K. – A girl of eight lives in a large provincial city. She was born in April of 1999 in a full family. The economic status of the family – average. She is brought up in a 3-room flat, rather small and cramped. She is the youngest child in the family. She has an older sister – 17 years old and an 11 year old brother. Her father is disabled, has a visual impairment due to pigmented retinopathy and is a graduate of a school for the visually impaired. The older sister similarly has a congenital visual impairment, and her vision is very poor. The brother is the only healthy child in the family. The mother has been suffering from diabetics for years (CVP IV); she does not work – looks after the children. The father has been a professional sportsman for 15 years – cycling with a guide. He has won the Polish, European and World championship for disabled people in tandem cycling, participated in Paralympic Games; at present he is actively preparing himself for the Paralympic Games in Beijing. He stays at home for longer periods of time only during three months of the year – in winter time. He devotes most of his time to intensive trainings. The mother is thus burdened with bringing the children up. The siblings are educated in an integrated school.

Malwina was born as the fourth child in the family. The first child, a boy, died shortly after birth. She was born at full term, according to the calculations performed on the basis of USG examinations conducted by the physician who supervised the pregnancy (two USG examinations were conducted – one at the beginning and one at the end of the pregnancy). However, the mother strongly believes the girl was a premature infant. The conviction results from difficulties in estimating the beginning of the pregnancy due to the mother's hormonal problems. The birth weight was 3030 gm; her Apgar scores were 7-8. Immediately after the birth foramen in the heart were diagnosed: interventricular (ASD) and interatrial (USD), which soon crusted over. However, Bottal's duct opened. In the first hours of her life, the child collapsed twice and was resuscitated. Only some days later, a cleft palate was noticed and consequently Pierre-Robin Sequence diagnosed. A child with Pierre-Robin Sequence is born with a small lower jaw moved backwards, which creates an impression that there is no chin; the basis of the oral cavity and its volume are smaller than usual, consequently the

tongue tends to fall back and downward towards the throat and so there is a danger of choking; cleft soft palate is a concurring anomaly. In the past the treatment involved a surgical intervention during which the tongue was moved towards the lower lip and fixed in a particular position. At present in the neonatal period and in the infancy preventive treatment is administered, with a prolonged endotracheal intubation (inserting into the trachea a tube made of rubber or plastic through the nose or mouth in order to ensure the patency of the respiratory tract), until the moment when the child acquires the swallowing and expectorate reflexes[6]. The analysed child was born without the sucking reflex. She was fed through a tube. She spent the first months of her life in the neonatal pathology department. For the first time she was brought home for a few days when she was two months old. She underwent numerous, sometimes shorter, sometimes longer hospitalisations both in her home town and in other cities in Poland. In time the following defects were diagnosed: severe hearing impairment of the sensory-nervous nature, above 90 dB, auditory meatus atresia, internal ear aplasia, pigmented retinopathy, myopic astigmatism of both eyes, hypothyroidism, cyst of the choroid plexus on the left lateral ventricle, valvular incompetence, narrow palpebral fissures, facial dystrophy, asymmetric body, better muscular tone on the right-hand side of the body, and hypoglycaemia. Until the 10th month of her life, Malwina had no muscular tone. At present, the visual impairment is being corrected by means of corrective glasses: the right eye – 7, visual area 105º, the left eye – 8, visual area 65º. Genetic examinations have been conducted, which so far have not diagnosed genetic defects. Karyotype 2N46 XX. Genetic examinations are being continued.

The child, which is understandable, was often treated with antibiotics. In 2003 she underwent an operation of the palpebral fissures in Instyt Matki i Dziecka (Mother and Child Institute). There she collapsed again. She turned blue in her mother's hands, who although terrified, as she claims, had no idea of what was happening. Only a nurse's intervention made her realize that her child's life was endangered. The resuscitation procedure was began immediately. Since the age of 5 Malwina has had a cochlear implant in her right ear. The implantation procedure was conducted in Instytut Fizjologii i Patologii Słuchu (Institute of Physiology and Pathology of Hearing). When she was 3 months old she began physical rehabilitation which lasted for 1.5 years. Minor progress in her motor development was noticeable as late as when she was some months old – she began to hold her head

[6] A. Wagner, Z. Kaliciński, *Chirurgia wieku rozwojowego*. In: *Encyklopedia zdrowia*, PWN. Warszawa 1994, p. 1700.

independently. However, her motor development – due to the influence of physical rehabilitation – did not follow existing standards. When she was 10 months old, Malwina was able to sit on her own for a short period of time when seated by her caretaker. She was unable, however, to reach the sitting position on her own or return to this position having moved to her back or side. When she was 1 year and 7 months old she started to walk, and when she was 2 she learnt to crawl on all fours, i.e., after she acquired the skill to walk.

Since she was 5 months old she has been included in the course of surdologopedic therapy in Ośrodek Diagnozy i Rehabilitacji Dzieci i Młodzieży z Wadą Słuchu Polskiego Związku Głuchych (Diagnosis and Rehabilitation Centre for Children and Youth with Hearing Impairment of the Polish Associations of the Deaf). She attended the therapeutic class once a week. The therapy was and still is conducted by the same specialist. Beginning at the age of 4 she participated three times in 5-week rehabilitation courses, organized for children with hearing impairment and cleft palate by Specjalistyczny Zakład Rehabilitacyjno-Leczniczy Narządów Mowy Zgromadzenia Sióstr św. Elżbiety w Otwocku (Speech Organs Specialist Rehabilitation and Treatment Institution of St. Elisabeth Female Order in Otwock). Unfortunately, the child's participation in such courses was connected with her separation from home and family. None of the close relatives – such were the rules – could accompany the child. The influence of the first two courses on the communicative skills, and most importantly on the interest in getting into relationships with other people – in the mother's opinion – was noticeably positive. The third course was evaluated by the mother negatively perhaps because during it the child had lymphadenitis and had to be hospitalised.

In the pre-school period Malwina attended a public nursery school. The aim was to develop communicative skills in interactions with healthy peers. Thus she was placed in a nursery class in an integrated school. She spent two years there including, as already mentioned, a one year delayed schooling. At present she can count to 20 using concrete objects. She can add and subtract up to 10. She knows the letters of the alphabet and can read simple texts. She paints and draws. She likes to dance and listen to children's music, plays with dolls, and gets involved in thematic games and watching cartoons. She has no problems with getting into interpersonal interactions with adults and children of her age using, generally, non-verbal components of communication, which are only sometimes verbalized. She is open to such contacts and gets actively involved in the communicative process. She experiences serious difficulties with articulated speech. Her speech is unclear, blurred, nasal and lacks proper pronunciation of almost all sounds. Sometimes the lack of comprehension of messages directed by the child to others results in her giving up on the continuation of the

interaction or a refusal to provide oral answers substituting for them non-verbal ones. She is a cheerful, smiling child, who appears to be happy.

On September 1, 2008 she began education in the first grade in an integrated school. Her mother spent the entire holiday period worrying whether the child would be able to cope with the educational curriculum for healthy children. Although there are serious doubts whether she will be able to compete with healthy peers, still, with the help of her family and people whom she has met in her life so far and who have provided her with support and understanding, she has definitely succeeded and this success is significant and indisputable. Yet initially the prognosis, as her mother recalls, was negative to such an extent that she had heard from one of the specialist that the child would be good for nothing, that she would be a vegetable till the end of her life and it would be better to place her in an institution. A notable consequence of such an approach was that on the discharge from the maternity ward, the mother was allowed only half of the maternity leave she was entitled to, which was motivated by the lack of hope for the child's survival. The mother was promised to have the leave prolonged in the event the child actually survived. As the future has shown, Malwina surprised all disbelievers and doubters and those who had given up on her.

Recalling the first days of Malwina's life, the girl's mother greatly regrets the lack of psychological help in that specific situation of giving birth to a disabled child, the lack of psycho-emotional support on the part of specialists, which according to her is very needed and might significantly soften the parents' emotions, especially mothers' emotions evoked by the described circumstances. She also notices the necessity of providing information concerning the child's health in a proper way, as well as describing possibilities to look for specialist assistance, specific institutions which support such children and actual perspectives concerning the child's development and the role of the family in this process. From the perspective of these 8 years, she can see that at present the medical care has improved significantly, the quality of which – as in the past – depends on the people who work in a health care system. Yet she still notices shortcomings, especially in the realms of the psychotherapeutic, informational and psychic support as well as the economic support of a family with a child with multiple disabilities. She ascribes the success of her child to being lucky in having met people who offered to help and actually did help by turning their words into actions. It is difficult to say what the future will hold for Malwina. Only time can answer this question. And one must hope that the answer will be satisfactory.

3. Conclusions

Any type of disability may complicate a human life and development in a unique way which is characteristic for this particular type. A person with a multiple disability, a disability which results from genetically-induced congenital anomalies, is faced with exceptional problems, which are sometimes similar and on other occasions completely different. Although for at least some years now specific actions have been taken to support such people medically, psychologically and pedagogically, still these activities are not satisfactory in correlation with those problems experienced by children, youth and adults as well as their families. Thus it is high time these issues were analysed in detail and for a proper system of providing special assistance to people who live with the aforementioned difficulties and their direct consequences to be devised.

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