CASE STUDY

ACTA Vol. 12, No. 1, 2014, 103-113
NEUROPSYCHOLOGICA

Received: 18.12.2013
Accepted: 28.03.2014

A – Study Design
B – Data Collection
C – Statistical Analysis
D – Data Interpretation
E – Manuscript Preparation
F – Literature Search
G – Funds Collection

SELECTED ELEMENTS OF THE PSYCHOSOCIAL FUNCTIONING OF A GIRL WITH HOLOPROSENCEPHALY

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SUMMARY

Background: The medial structures of the brain play a key role in the integration of all environmental stimuli that become the object of conscious awareness. Their integrational functioning is a factor not only of the position of these structures, but also their anatomical structure. They provide communication and integration between the lower and higher functional levels of the central nervous system.

Case study: Persons suffering from holoprosencephaly (HPE), then, can have difficulties with perception, operationalization, and organization in general. As a result, the dynamics of their psychosocial development can take a very specific form. We present excerpts from nine years of observation over the course of rehabilitation of a girl with the middle interhemispheric variant (MIHV) of HPE. This was a specific, exceptional, and unique course of development, and our encounter with this child in the course of rehabilitation allowed us to observe some very particular phenomena regarding her individual development in the domains of motor and sensory functions, communication and language, emotional development, and contact with people.

Conclusions: Compensation of developmental deficits is possible and complete, if early support for the child can be commenced, as early as possible, virtually from the first moments of life, and certainly after an accurate diagnosis has been reached. This is how the proper course of rehabilitation should run.

Key words: neurodevelopmental disturbances, cognitive functions, perception, action, compensation

INTRODUCTION

Holoprosencephaly (HPE) is a complex congenital malformation of the brain, the essence of which is an incomplete separation of the cerebral hemispheres (Golden, 1999). The process of lateral expansion of the hemispheres from the kernel of the forebrain is completed at the beginning of the second month of fetal age; possible disturbances in the course of this process produce abnormalities in the structure of both the brain and the face (Bochenek & Reicher, 1993; Golden, 1999). Matsunaga and Shiota (1977) found that HPE occurs in one of every 250 pregnancies, but only 3% of these children survive the post-natal period (Cohen, 1989). Various authors have estimated the frequency of occurrence of HPE among all children brought to term and born as 1 in 10,000 (Croen et al., 1996; Rasmussen et al., 1996; Bullen et al., 2001), 5-12 in 100,000 (Roesler et al., 2001), or 0.56-0.63 in 10,000 (Niknejadi et al., 2008). Orioli and Castilla (2010) performed a broad ranging analysis of materials containing epidemiological data, and created a complex and very interesting picture of HPE, not only in terms of the frequency of this phenomenon, but also the risk factors for its development.

The term holoprosencephaly was proposed by DeMyer and Zeman in 1963, beginning a long and complicated road of description and explanation of the genesis of certain congenital, developmental malformations of the brain. They also suggested a classification of HPE, distinguishing three forms: alobar, semilobar, and lobar. This is not the only classification of HPE, and particular attention should be drawn to two others, constructed respectively by Probsta (1979) and Leech and Shuman (1986).

Forms of HPE

The point of reference for distinguishing forms of HPE are the brain structures located along the medial line, including those located deeper, on the level of the midbrain.

Alobar

- complete or almost complete lack of separation between the left and right cerebral hemispheres;
- a single ventricle along the central line in the front;
- absence of the central fissure;
- misshapen olfactory bulbs, corpus callosum, and falx;
- undifferentiated subcortical gray matter bodies.

Semilobar

- non-separated hemispheres in the front, with visible separation in the posterior segment;
- lack of the anterior cornu of the lateral ventricles, the septum pellucidum, and the anterior part of the corpus callosum;
- absent or hypoplastic olfactory bulbs;
- incompletely separated subcortical gray matter bodies;
- possible dorsal cyst.

Lobar

- well developed central fissure;
- incompletely developed subcortical structures;
- dimorphicity of the ventricles, with a well-formed third ventricle;
- hypoplasia of the falx and the olfactory bulbs;
- absence of dorsal cyst.

Children with HPE also manifest phenotypic diversity, which often makes it possible to hypothesize a diagnosis even prior to performing an MRI (Solomon et al., 2010; Richieri-Costa & Ribeiro, 2006).

The diverse clinical pictures presented by the brains of children with HPE has inclined many researchers (including Barkovich & Quint, 1993; Simon, 2002; Solomon et al., 2010) to supplement DeMyer and Zeman's classic division with a fourth type, the middle interhemispheric variant (MIHV). The degree of malformation of the structures of the medial forebrain in a child with MIHV is considerably less than in the classic forms of HPE described above. This form is characterized by a mild course and the possibility of far-reaching therapeutic interventions in rehabilitation.

All forms of HPE can be placed on a continuum in terms of the degree of malformation of brain structures and the corresponding functional difficulties (Solomon et al. 2010). Hahn and Plawner have shown that abnormal muscle tension and problems with locomotion are present in all forms of HPE. The type of HPE is also significantly differentiated by the degree of motor dysfunction. In this group, patients with MIHV display the highest level of overall functioning, despite observable disturbances, such as hypotonia, dystonia, or spasticity. The absence of involuntary movements in MIHV makes it much easier for these pa-

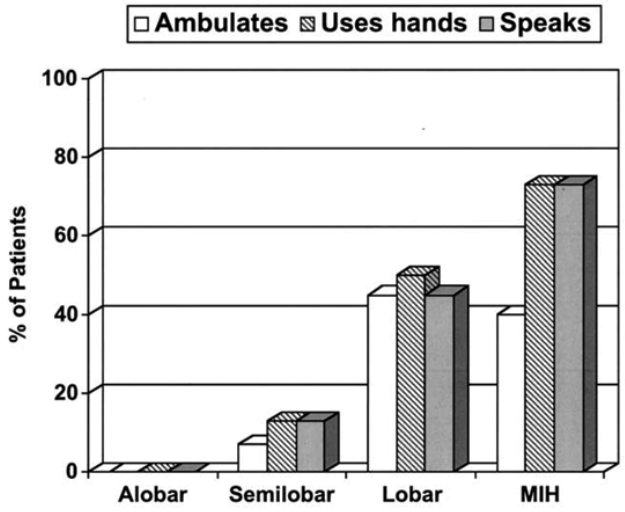

Fig. 1. Correlation between the type of HPE and the development of ambulation, hand movements, and expressive speech (Hahn & Plawner, 2010)

tients to achieve better quality of life, as compared to patients with other forms of HPE. Hahn and Plawner (2010) have also called attention to associations between the forms of HPE and the development of walking, hand movements, and expressive speech (Fig. 1).

CASE DESCRIPTION

Agnes, the patient to be described, was born in 2003 with MIHV. Since the very beginning of her life, she has needed to be under the constant care of physicians, especially neurologists. Rather early on, she was found to have epilepsy. Several months later, an MRI examination of the structural status of her brain showed fusion of the thalamus. In the images of the brain, large thalami were observed, which were widely connected with each other, as result of which the opening of the third ventricle was not visible. This examination also showed that the shape and structure of the corpus callosum was normal, and the gyri of the brain were well developed. Another MRI done in 2008 clearly confirmed the presence of changes in brain structure characteristic for MIHV. On the neuronal level, the examination revealed heterotrophy of the gray matter, and no interference in the flow of signals through the white matter. Agnes's visit to the Early Intervention Center was the beginning of our long-term observation, and she began psychological rehabilitation.

Agnes's psychomotor development had a very uneven course, as shown in parts of Table 1. Between the ages of two and three, she began meeting with therapists in the Early Intervention Center. The process of rehabilitation was directed holistically towards all domains of functioning. The integration of medical treatment, rehabilitation, and psychological help made it possible to describe certain psychological events, the appearance of which showed a unique and individual line of development, extending into the future.

Since our first meeting, Agnes has surprised us with her unique and unexampled picture of development. In analyzing over time the course of her development in the area of communication and language, we have observed that her skills in this respect appear in clusters. Between the ages of two and three, she discovered and pronounced her first word (recognizable as the Polish word for "where"), and what she could not pronounce, she replaced with gestures. Some of her gestures and their interpretations can be found in Table 2. All of these gestures had a very concrete reference, rooted in situations of daily life. The discovery of these gestures and the support of her family made it possible for her to become more communicative, and she entered into relationships with other people with less anxiety. During this same time, Agnes began to use a pointing gesture; however, she did this, not with her index finger, but with her thumb. The impossibility of communicating effectively with the index finger caused it to be replaced for this purpose by another finger, over which she had greater control. Since that time she has been able to participate in communication, in both the perlocutionary and illocutionary modes (Austin, 1962). Thus it became possible

for her to take the initiative in communicating with other people. Pointing with her thumb continued until she was about five years old, after which time the pointing function was taken over by the index finger. In analyzing the evolution of this gesture, we had the impression that the most important factors in this process were her parents' openness and her unimpaired communicative resources in the

Table 1. Selected aspects of the patient's development (moment of appearance) in chronological order

		Developmental age								
Aspect of development		0-1	1-2	2-3	3-4	4-5	5-6	6-7	7-8	8-9
M/S	Agnes begins to walk; gait is very unsteady.		x							
	Hypersensitivity of the hand diminishes, Agnes begins to touch and grasp different objects and substances, and supports herself on both hands when she stands up from the floor.				x					
	Agnes no longer avoids place with uneven floors when walking.								x	
C/L	Agnes says her first word (*dzie,* for Polish *gdzie* 'where').			x						
	Gestures are the primarily communicative tools.			x						
	Agnes uses the thumb instead of the index finger to point.						x			
	Agnes says more and more words, enriches her vocabulary.						x			
	Grammatical compensation appears in Agnes's utterances ("dry not" means "wet").								x	
	Agnes speaks in sentences and can be understood even by people who d not know her.								x	
	Agnes communicates all her needs.								x	
E	Agnes is no longer afraid of strangers; she exercises in rehabilitation and allows the doctor to examine her.						x			
S	Agnes begins to cooperate with other people.						x			

Explanation of abbreviations: M/S - motor and sensory, C/L – communication and language, E – emotional development, S – contacts with people

non-motor aspect. The need to communicate, and as a result the formation of adequate relations with important people as a goal, caused the choice of whatever means in a given moment guaranteed success in communication.

Between the ages of five and six, Agnes was saying more and more words, constructing their form according to some very interesting, "innovative" rules. Samples are given in Table 3.

Agnes's active vocabulary has undergone major changes, especially in respect to the abstract-concrete attitude. Alongside words with very concrete meanings, there were also more abstract words and words referring to time. The expression *ha-siu* meant "tomorrow" for her, but initially for Agnes this was the moment when she woke up. The sequence of sounds *siora* has a phonetic similarity to the Polish word *wczoraj*, which means "yesterday." Her family discovered these moments in thinking about and experiencing time, gradually reinforcing and rewarding her for correct usage of these expressions. Additional words associated with time were realized in a rhythmic single and double repetition of the basic words (*ha-siu* and *siora*). Between the ages of seven and eight there was a further qualitative change, when syntactic structures appeared. Just as concrete words prevailed in the case of vocabulary, so her sentences contained ref-

Table 2. Selected gestures and their explanation

Appearance of the gesture	Meaning
wiping her hands together	butter her bread
a diagonal movement of the hand, repeated several times, at the level of the trunk	slice her bread
energetic and simultaneous lifting both arms	throw a ball
rapid movement of the trunk backwards and forwards	go on the swing
rapid and short movement of the open hand up and down at mouth level	wipe her face

Source: Mański, 2010.

Table 3. Selected elements from Agnes's active vocabulary

verbalization	references/denotations of Agnes's words
ja, tata, baba, ma, asia, ciocia, nie	Ja (**I**), Tata (Dad), Babcia (Grandma), Mama, Asia (her sister), Aunt, no [all these words are correct in Polish]
mm, mu, zi, chch, dzia	car, cow, cold (*zimno* in Polish), warm, Grandpa (*dziadek* in Polish)
ha-si;, ha-siu ha-siu;	tomorrow, day after tomorrow
siora; siora siora	yesterday; sometime

Source: Mański, 2010.

erences that for the most part were directly related to situations and persons close to her. Despite her limited active vocabulary, she could construct messages using the resources she possessed. She displayed extraordinary ingenuity, and thanks to the quick reactions of her parents, every signal containing even a slight communicative meaning was reinforced. This can be illustrated by several examples:

- Agnes learned and properly used the phrase “Auntie Eee”, which referred to her aunt Irena. A necessary condition for the appearance of anyone’s name in her active vocabulary was a close personal bond with that person. When she tried to learn also the role and name of the spouse of her aunt Irena, she produced the form “Unk Eee,” that is, Uncle Maciek, Irena’s husband.
- Agnes learned and properly used the word “dry” to specify the lack of water in an object. After showing her a sponge that had not yet been wetted, we asked her to describe the sponge and received a correct response. In order to specify the state opposite to dryness, she invented the expression “dry not”.
- The verbal negation of certain activities evolved according to a very precise model. When she did not want to go to some place, she first used only the word “no”, then the structure “go no”, and finally the form “does not go”. We observed a similar course in the case of washing. The developmental sequence went from “no” to “wash no” to “not wash”.

Agnes’s efforts to communicate, despite significant motor limitations, were very quickly noticed first by her family, who later assisted specialists in understanding that she is a fully intentional child. At around the age of six, a breakthrough took place in her reaction to persons outside her family. She was no longer afraid of strangers, and was more trusting towards psychologists, physical therapists, and other therapeutic personnel, which made it possible to expand the range of interventions. This greater trust in strangers, combined with a readiness to cooperate with them, made it possible for her to find a place in an integrated preschool. Thanks to this, Agnes began formal education.

The last aspect of development that will be described here is the sensory-motor domain. Against the background of the whole of Agnes’s development, motor functions were the first to become a target for rehabilitation efforts. She began to walk between the ages of one and two. She showed a definite preference for flat surfaces. Every unevenness in the floor caused her to lose her balance, or she simply refused to walk. In order for her to walk without impairment, it was particularly important that she have confidence in the architectonic characteristics of the place. Not every fall triggered a supporting reaction, and so they became especially dangerous. In her dynamic profile while walking she was perceived to lean forward (even in standing position) and to make incomplete use of her entire foot, as though the floor were covered with “glowing coals” over which it was necessary to pass very quickly.

In the course of therapy taking place in space, Agnes showed her therapists an extraordinary feature qualitatively associated with planning movement. In tasks requiring her to cross over a path leading to a target and then returning to

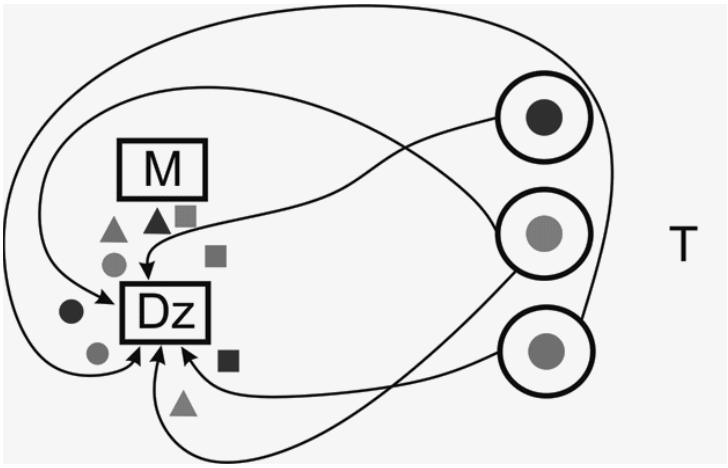

Fig. 2. Scheme of paths of return used by Agnes in exercises. Key: M – where Mother is standing; Dz – where Agnes is standing ; T – where the therapist is standing. Source: Mański, 2010.

a previously established place, she showed extraordinary diversity in her return paths. Each time, she returned to the therapist or the chosen target by another path. The goal remained unchanged in space (e.g. a chair, a box, a table), while the trajectories of movement did not coincide, even when that would have provided economy of movement (the shortest path). She constantly corrected the shape and direction of her path. Figure 2 presents a typical task, in which it was necessary to cross over space to find the appropriate loop and put a figure in it. The lines between the collection of loops and the boxes are examples of her "trajectories of return." The dotted line showing the shortest route was used during the sessions as often as the remaining, longer paths. It was as though each time she constructed space from the beginning and learned the path, except that the learning was ineffective.

DISCUSSION

Solomon et al. (2010), in characterizing MIHV, have pointed to the rather frequent occurrence in this form of HPE of incomplete separation of the thalamus. From a neuropsychological perspective, the thalamus, integrating signals from receptors on the skin, internal organs, and the senses of sight and hearing, is the most important collection point connecting the lower levels of the nervous system with the higher. Duus (1989) called attention to the fact that in order for any signal to be consciously received, it must past through the thalamus, which has been accurately named "the gateway to consciousness" (Duus, 1989). The material presented here, along with the neuropsychological reference, has many practical implications associated with multi-dimensional rehabilitation. Table 4 presents selected aspects of Agnes's behavior, along with a neuropsychological explanation.

Table 4. Neurological approach to selected aspects of Agnes's functioning

Aspects of functioning	Neuropsychological premises
Agnes does not touch snow, wet grass, or warm sand on the beach…	In the thalamus the basic sensations, such as pain, cold, heat, warmth, dryness, etc., become objects of conscious awareness.
Agnes prefers flat surfaces, will not enter a place with an uneven floor.	The thalamus mediates in the sending of signals from the hypothalamus, brainstem, and cerebellum to cortex, which is why it is important for locomotor processes.
Agnes diversifies her return paths during exercises that require moving through space.	The double connections between the thalamus and the brain condition the modifying effect of the motor cortex on motor functions.
Agnes avoids touching or grasping liquids of various consistencies, such as modeling clay, paint, glue, etc.	The thalamus gives a particular affective values to various afferent impulses, and transmits this information to the higher levels of the CNS, which causes a positive or negative attitude towards the stimulus.
Agnes does not know how to turn her head to shield her face from wind or rain.	The thalamus, in connection with the extrapyramical system, is a coordinating center, and plays a major role in the genesis of movements in response to pain or other sensory or affective stimuli.

In our opinion, the success of early therapeutic interventions improving quality of life of both Agnes and her family may have their source primarily in the baseline state of the medial structures. The status of these structures should be taken into account by the therapeutic team in planning comprehensive rehabilitation. The authors of the present study would like to call attention to two hypotheses – "lack of structure" and "malformed structure" - which can explain the conditions and course of the process of integration, depending on the extent to which a given brain structure actually exists. A particular structure (e.g. the corpus callosum) may be completely absent, a state which we call agenesis of the corpus callosum. In the second case, the structure may be malformed, or its structure may be changed. Examples of this would include dysgenesis of the corpus callosum or fusion affecting paired structures (e.g. the thalamus). When a structure does not exist, signals and stimuli pass around the place of the missing structure, forming circular paths based on other structures. The compensation can be complete, if we begin early support from the very beginning of the child's life. When a structure exists in a malformed condition, the signals are processed by it, but the effect of the integration of signals can be a change that is insignificant from the point of view of higher levels of the nervous system, and does not become the object of consciousness.

CONCLUSIONS

The phenomena and more important developmental achievements described here chronologically show that Agnes's quality of life was directly conditioned by her motor functions, communicative competence, and emotional maturity, enabling her to exist in social relations. The effect of walking was achieved to

a large extent thanks to the early introduction of rehabilitation. Agnes crossed even very large distances with passion and enormous involvement. The people around her did not concentrate on the formal correctness of her walking, but rather on the great joy that Agnes experienced when she could take a walk with someone. Her motor development led to curiosity about the world and people, but even earlier her orientation towards people was very obvious.

The next sphere that improved her quality of life was communication. Her desire to communicate with her family was so strong that even her seriously limited articulatory resources did not keep her from displaying grammatical competence. The development of communication enabled Agnes to attend preschool, and then a kindergarten with a integrational profile. The increase in her communicativeness was brought about by intensive educational activities and speech therapy, but even more by her excellent communicative relations with her family. She grew up from the beginning in an open society; she had contacts with children, and she participated in the life of both her closest and farther families. Adaptation in these relationships made it possible for Agnes to discover cooperation with her family and peers. All the aspects of development presented here, as it is showed in the syndromological analysis (Pachalska & Kaczmarek), affected the outcome and her quality of life (see also: Pachalska 2009). If not for the high level of health care provided by the parents and the multi-dimensional specialized help, keyed to the schedule of developmental events, she would be far more dependent on her family and her life would be governed only by her deteriorating health.

Conclusions: Work with Agnes and her parents, who provided the first rehabilitation, once again confirms the thesis advanced by the authors of the present study, that compensation of developmental deficits is possible and complete, if early support for the child can be commenced, as early as possible, virtually from the first moments of life, and certainly after an accurate diagnosis has been reached. This is how the proper course of rehabilitation should run.

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