Dandy–Walker Syndrome in Poland: Review of Patient Health Outcomes

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Abstract

Dandy-Walker syndrome is a congenital defect of a central nervous system characterized by a hindered flow of cerebrospinal fluid through the outflow holes of ventricle IV. Symptoms of the disease can develop slowly since the early childhood and will result in developing both hydrocephalus and anomalies related to the structure of the cerebellum. The etiology of Dandy-Walker syndrome is not known.

Aim of this work is to present the health situation of the patients with Dandy-Walker syndrome in Poland.

Material and methods

The research was conduncted from December 2011 to April 2012 and involved the group of wards of the ‘Podaj Dalej’ Dandy-Walker Syndrome Foundation who live on the territory of Poland. Overall, 36 people were examined: 15 boys and 21 girls.

Results and conclusions

Reaching the patients with Dandy-Walker syndrome is very difficult because it is a rarely occurring congenital defect, recognized usually by accident. However, the complexity of problems evolving from this disease brings the necessity of expanding the studies in many areas so they could be compared to the studies conducted in other countries. Results of this type of studies will be useful in a correct orientation of treatment and widely understood rehabilitation of patients with Dandy-Walker syndrome as well as education both families and the patients.

Introduction

Dandy-Walker syndrome is a congenital defect of a central nervous system characterized by a hindered flow of cerebrospinal fluid through the outflow holes of ventricle IV. Development of the disease takes place probably in the first trimester of pregnancy when due to disorders of circulation or infection the structural damages appear [1]. I used the word 'probably' because nowhere in the medical literature there is assurance that the process of this syndrome is carried out exactly in this way. The fact is however, that the symptoms of this rare disorder can take place both in childhood and in a mature age. The characteristics of Dandy-Walker syndrome are:

- dysplasia of the cerebellar vermis,
- higher location of the tentorium cerebelli
- cerebellar hemispheres moved to the sides and usually hypoplastic
- partial or complete closure of the IV ventricle holes
- arachnoid mater cyst in the roof of fourth ventricle

As the consequence of such disturbed architecture of posterior cranial fossa, the hydrocephalus in 53-82% of cases has developed [4,8,11].

Symptoms of the disease can develop slowly since the early childhood and will result in developing both hydrocephalus and anomalies related to the structure of the cerebellum.

Frequency of occurrence of this syndrome is estimated in literature to be 1:25,000 – 1:35,000 although the authors agree that the frequency of detection of the disease could improve in the times of standard ultrasonography performed on pregnant women [8,9]. The etiology of Dandy-Walker syndrome is not known. In the literature we find reports on the datum pointing to the leading role of genetic factors in the form of chromosomal pathology, especially trisomy 18, trisomy 13, Turner syndrome, tetrasomy 9p and 9pqh coexisting with a karyotype 47 XY + dic (9) (q12) in peripheral blood lymphocytes and with a normal karyotype marked with fibroblasts [3,5,6].

Aim of this work is to present the health situation of the patients with Dandy-Walker syndrome in Poland.

Material and methods

The research was conducted from December 2011 to April 2012 and involved the group of wards of the 'Podaj Dalej' Dandy-Walker Syndrome Foundation who live on the territory of Poland as well as new persons applying at that time for consultation to the doctors supporting activity of Foundation. Overall, 36 people were examined: 15 boys and 21 girls.

Parents of ill children answered the questionnaire prepared especially for the needs of the research in the electronic or paper form at their homes after having agreed for

conducting the studies, and later they sent the answered questionnaires by electronic or traditional post.

Results

Identification of Dandy-Walker syndrome in the studied group proceeded differently and usually was accidental during the examinations directed at detecting another disease. In 7 cases, the DWS (Dandy-Walker syndrome) was identified in fetal life, 15 in first year of life and 14 in the subsequent years.

Distribution of characteristics of Dandy-Walker syndrome in the case of studied group was presented in the Table I and is as follows:

1. Changes regarding the cerebellar vermis – 75% of the respondents
2. Cyst communicating with the fourth ventricle – 58.3% of the respondents
3. Changes regarding cerebellar hemispheres – 25% of the respondents
4. Enlargements of spinal-cerebellar tank – 38.8% of the respondents

Table I Distribution of characteristics of Dandy-Walker syndrome in the studied group

Answers	Studied group	
	N	%
Lack of cerebellar vermis	4	11,11
Cerebellar hemispheres widely spaced	5	13,89
Small, hypoplastic cerebellar hemispheres	4	11,11
Hypoplasia of the cerebellar vermis	23	63,89
Normal cerebellum	0	0,00
Enlargement of the spinal-cerebellar tank	14	38,89
Arachnoid mater cyst communicating with the fourth ventricle	21	58,33
Other	8	22,23

Nine children have an intraventricular shunt installed. Among this group, the following complications took place: shunt system obstruction, over-draining, shunt

system infection, peritonitis and catheter movement outside the peritoneum. Summary of these complications is presented in Table II:

Table II Summary of complications after intraventricular shunt implantation in the studied group

Answers	Studied group	
	N	%
Obstruction	7	19,44
Over-draining	2	5,56
Shunt system infection	1	2,78
Peritonitis	0	0,00
Catheter movement outside the peritoneum	2	5,56
Other	1	2,78
Overall	11	30,56

Genetic studies were conducted among 15 from 36 families. In 10 cases, the necessity for such studies came from the suspicion of genetic disease coexisting with Dandy-Walker syndrome. In other 5 cases, the studies were conducted on the explicit request of parents. The results are presented in Table III.

Table III Results of genetic studies in the researched group

Answers	Studied group	
	N	%
Trisomy 18	2	5,56
Trisomy 13	1	2,78
Turner syndrome	0	0,00
Tetrasomy 9p i 9qh	0	0,00
Karyotype 47XY + dic(9)(q12) in peripheral blood lymphocytes	1	2,78

Answers	Studied group	
	N	%
Normal karyotype marked with fibroblasts	10	27,78
Trisomy 9p	0	0,00
Other	3	8,33
Overall	36	100,00

In 10 cases, the karyotype marked with fibroblasts was correct. 2 people have trisomy 18 and in singular cases the trisomy 13 and karyotype 47XY+dic(9)(q12) in peripheral blood lymphocytes were identified.

At all the patients included in the examination, the coexistence of different defects and diseases was found. At the forefront is mental underdevelopment occurring in 30.5% of the respondents. 27.7% respondents are under the constant care of ophthalmologist due to amblyopia and in 9 cases there was a need to correct strabismus. 13.8% of the respondents have spastic paraplegia and the same number of patients has a diagnosed defect of the interventricular septum. The singular cases present cystic renal dysplasia, autism and Arnold-Chiari syndrome. 6 children have pathological changes of face and limbs and the same number of cases is for microcephaly. In an overview of coexisting defects and diseases presented in the Table IV there is also a column ‘other’. This lists defects and diseases that are not included in the literature as those coexisting with Dandy-Walker syndrome, but they have occurred in the case of the studied group. They include:

- Klippel-Feil syndrome,

- Ehlers-Danlos syndrome,

- Cryptorchidism,

-Hypoacusis,

-Atrophy of the optic nerve.

Table IV Defects and diseases coexisting with Dandy-Walker syndrome occurring in the studied group

Answers	Studied group	
	N	%
Mental underdevelopment	11	30,56
Spastic paraplegia	5	13,89
Diaphragmatic hernia	2	5,56
Defect of the interventricular septum	5	13,89
Cystic renal dysplasia	1	2,78
Pathological changes in face and limbs	6	16,67
Autism	1	2,78
Strabismus	9	25,00
Amblyopia	10	27,78
Microcephaly	6	16,67
Arnold-Chiari syndrome	1	2,78
Other	16	44,44
Overall	36	100,00

As one of the main problems, parents of children with Dandy-Walker syndrome indicate changes in the children's behaviour characterized by aggression (27.7%), depressed mood states (27.7%), sudden and frequent changes of mood (44.4%), irritability (47.2%). This is a serious problem for guardians, because it brings difficulties in upbringing and the reason for such states is not always known. Another difficulty particularly severe for the families of children with Dandy-Walker syndrome and indicated by all the respondents is the lack of information about the disease itself and its consequences. Hence, the rehabilitation of children is often undertaken with delay and this brings other problems.

Literature discussion

For the question about conducted genetic studies 41.6% of the respondents answered positively. The results presented in Table III can be compared to the research conducted in other countries. The table includes given by literature chromosomal pathologies which coexist with Dandy-Walker syndrome. Some of them exist in the case of the studied group.

In contrast, no one has been found with a trisomy 9p ust 9 ptr → p11.2, which, as the studies published in the journal 'Prenatal Diagnosis' show, may be the cause of Dandy-Walker syndrome [2]. The slight predominance of female fetuses with Dandy-Walker syndrome can be noticed. In literature this ratio is given as 3:1 [7]. In case of my studies this difference is smaller: females represent 58.3% of respondents.

According to the studies conducted in the USA [8,9], the morphological characteristics of Dandy-Walker syndrome appearing in the ultrasonography picture can be divided into three groups. In 50% of cases there is a lack of cerebellar vermis; 30% have a vermis defect and 20% of cases have small hypoplastic cerebellar hemispheres. In case of studies conducted in Poland this ratio is completely different. Persons with lack of cerebellar vermis represent only 11.11% of the whole group and they also have small hypoplastic cerebellar hemispheres. However, the vermis defect was found at 63.89% of the respondents.

Table IV shows comparison of diseases and defects coexisting with Dandy-Walker syndrome and occurring at the respondents. Such picture of the population examined matches the reports from the English literature. For instance, it is described that 40-80% of children with Dandy-Walker syndrome who have different types of somatic pathologies demonstrate characteristics of mental underdevelopment at the late stage (IQ below 70) [8,9,10]. In case of the group studied, 30.56% children are in such situation.

Conclusions

Reaching the patients with Dandy-Walker syndrome is very difficult because it is a rarely occurring congenital defect, recognized usually by accident. However, the complexity of problems evolving from this disease brings the necessity of expanding the studies in many areas so they could be compared to the studies conducted in other countries. Results of this type of studies will be useful in a correct orientation of treatment and widely understood rehabilitation of patients with Dandy-Walker syndrome as well as education both families and the patients.

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