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ARTIGO ORIGINAL

Perfil dos Pacientes com Mielomeningocele da Associação de Assistência à Criança Deficiente (AACD) em São Paulo – SP, Brasil

Profile of the Patients with Myelomeningocele from the Associação de Assistência à Criança Deficiente (AACD) in São Paulo – SP, Brazil

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RESUMO

Introdução: Dentre os defeitos de fechamento do tubo neural a Mielomeningocele (MMC) é a mais freqüente (85%). A etiologia é desconhecida, mas com características genéticas e ambientais. O diagnóstico pode ser feito no período pré-natal através de ultra-sonografia morfológica. Recomenda-se o fechamento da bolsa nas primeiras horas de vida e derivação ventrículo peritoneal (DVP) precoce. Objetivo: Traçar o perfil dos pacientes atendidos na Clínica de MMC da AACD - SP e pontuar as condições em que estes chegam à instituição objetivando verificar se ocorre o diagnóstico precoce e aprimorar as condutas do tratamento. Método: Revisão em prontuários de pacientes atendidos em avaliação inicial na Clínica de MMC da AACD - SP durante o ano de 2000, com idade inferior a um ano. As informações foram obtidas dos prontuários através de um protocolo de pesquisa. Entre as informações colhidas tem-se: dados pessoais, nível neurológico na primeira consulta, diagnóstico pré-natal, idade de fechamento da bolsa, presença ou não de DVP. Resultados: No total passaram 230 pacientes em avaliação inicial no ano de 2000. Destes, 64 (27%) apresentavam menos de 1 ano de idade na primeira consulta. A média de idade na avaliação inicial para estes pacientes foi de 5 meses. Destes, 44% eram do sexo feminino e 56% do sexo masculino. Em 37% dos pacientes o diagnóstico não foi feito no período pré-natal. Ao analisarmos a idade de fechamento da bolsa temos que em 51% dos pacientes isto ocorreu nas primeiras 24 horas de vida. Somente 17% dos pacientes não tinham sido submetidos à DVP até o momento da avaliação inicial. Ao analisarmos o nível neurológico na avaliação inicial observamos que 35% são do nível Torácico, 29% do nível Lombar Alto, 24% do nível Lombar Baixo, 11% do nível Assimétrico e nenhum paciente de nível sacral. Conclusão: Por ser a AACD - SP centro de referência no tratamento de MMC pode ser explicado o fato de recebermos maior número de crianças com níveis funcionais mais altos. É importante tentar estimular tanto a prevenção como o diagnóstico e tratamento precoce desta patologia visando diminuir o impacto que esta causa na sociedade. Foi achado esperado o não diagnóstico ou o diagnóstico tardio de MMC em nosso estudo, pois em nosso país ainda não é realizado de forma rotineira nos postos de saúde o ultra-som morfológico durante o acompanhamento pré-natal da gestante. A sobrevida dos pacientes com MMC tem aumentado devido ao fechamento precoce da bolsa e controle da hidrocefalia com DVP, associado posteriormente ao controle da bexiga neurogênica.

PALAVRAS-CHAVE

criança, mielomeningocele, hidrocefalia, reabilitação, centros de reabilitação

ABSTRACT

Introduction: Myelomeningocele (MMC) is the most frequent of the Neural Tube Defects, with 85% of the cases. The etiology is unknown, but it has genetic and environmental characteristics. The diagnosis can be achieved in the prenatal period through the morphological ultrasonography. The closing of the defect within the first hours after birth is recommended as well as the early control of hydrocephalus. Objective: To describe the profile of the patients treated at the Myelomeningocele Clinic of the Assistance Association to the Defective Child - AACD – SP and assess their clinical condition upon admission at the Institution, aiming at verifying whether the early diagnosis has been attained and improving treatment procedures. Methods: To review the files of patients seen at the initial assessment of the MMC Clinic of AACD – SP during the year of 2000, aged younger than one year. The information was obtained from patients' files through a research protocol and consisted of personal data, neurological level at the first assessment, prenatal diagnosis, age at the closing of the defect and presence or absence of ventriculoperitoneal shunt (VPS). Results: A total of 230 patients were assessed in the year 2000. Of these, 64 (27%) were younger than 1 year at the initial assessment. Mean age at the initial evaluation was 5 months and

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44% of the patients were females, whereas 56% were males. Diagnosis at the prenatal period had not been achieved in 37% of the patients. The closing of the defect was performed within the 24 hours after birth in 51% of the patients. Only 17% of the patients had not undergone VPS at the time of the initial assessment. The neurological assessment at the initial evaluation disclosed 35% thoracic, 29% high lumbar, 24% low lumbar, 11% asymmetric and no patient at the sacral level. Conclusion: The AACD - SP is a Reference Center in the treatment of MMC and this might account for the fact that the center receives a large number of children with higher functional levels. It is important to try to stimulate not only the prevention, but also the diagnosis and early treatment of this pathology, aiming at decreasing its impact on society. The lack of or late diagnosis of MMC was an expected finding in the present study, as the morphological ultrasound is not routinely performed in Basic Health Units in our country during the prenatal follow-up. The survival of patients with MMC has increased due to the early closing of the defect and hydrocephalus control through VPS, associated with posterior control of the neurogenic bladder.

KEYWORDS

child, meningomyelocele, hydrocephalus, rehabilitation, rehabilitation centers

INTRODUCTION

Myelomeningocele (MMC) is the most frequent of the neural tube defects, affecting 85% of the total number of cases.[1] A sac covered by epidermis can be seen externally, containing, internally, the spinal cord and nervous roots, both dysplasic, surrounded by cerebrospinal fluid (CSF). The defect occurs between the third and fifth weeks of intrauterine life, due to the failure of fusion of the posterior elements of the vertebral column.

The dietary supplementation with folic acid (vitamin B9) for child-bearing age women for at least three months prior to conception is considered to be effective in the prevention of MMC.[1]

The MMC etiology is unknown, but it has multifactorial characteristics (genetic and environmental). It is known that women with low dietary intake of folic acid present a higher chance of having children with MMC. The world incidence is variable, with an average of 1/1000 live births. The chance of recurrence is 5% for a second child, 10% for a third child and 25% for a fourth child of the same couple.[1]

The diagnosis can be attained in the prenatal period through the morphological ultrasonography (where a widening of the vertebral canal can be observed), the alpha-fetoprotein (AFP) measurement in the mother's blood (which is not specific, but of which elevated levels strongly suggest the disease) and acetylcholinesterase electrophoresis in the amniotic fluid.

The closing of the sac (to cover the exposed nervous tissue) is recommended within the first hours after birth, if possible before the first breastfeeding in order to prevent intestinal colonization by the bacteria present in the milk. The early ventriculoperitoneal shunt (VPS) is also recommended, to prevent the worsening of the neurological picture, which can lead to cognitive alterations, as well as to reduce the occurrence of cerebral herniation – known as Chiari Malformation Type II.[2]

The clinical picture is characterized by flaccid paraplegia and sensitive alteration below the injury (which can course with pressure ulcers, perforating plantar ulcers or burns), accompanied by neurological (hydrocephalus, tethered cord), urological (repeated UTIs, incontinence, calculi, reflux), and orthopedic impairment (trunk and lower limb deformities). Obesity can affect up to 40% of the cases.[3] Sleep apnea can occur in up to 20% of the cases[4] and latex allergy in up to 30% of the cases,[1] making it necessary to establish a "latex-free" environment for these patients.[5]

Hoffer classified MMC in functional levels according to the neurological impairment: thoracic, high lumbar, low lumbar, sacral and asymmetric.[6] The thoracic level doe not present active mobilization of the lower limbs. The high lumbar level has functioning psoas, adductors and, eventually, the quadriceps muscles. The low lumbar level has functioning psoas, adductors, quadriceps, medial knee flexors (hamstrings) and, eventually, anterior tibial and/or gluteus medius muscles. The sacral level has the aforementioned functioning muscles as well as plantar flexion and/or hip extension functions.

OBJECTIVE

The objective of the present study is to describe the epidemiological profile of patients treated at the MMC Clinic of AACD – SP and assess their clinical conditions upon their arrival at the institution, aiming at verifying whether the early diagnosis of MMC, as well as its possible complications, has been attained in order to improve treatment procedures.

MATERIAL AND METHODS

This is a retrospective study of the patients treated at the MMC Clinic of AACD – SP, based on the information obtained from the patients' files through a research protocol. Among the information obtained are the patients' personal data, prenatal diagnosis, pregnancy information and age at the closing of the defect as well as the presence or absence of associated diseases. The neurological level at the initial evaluation followed the classification proposed by Hoffer;[6] however, those cases that presented functional level asymmetry were also considered.

All files of patients who were initially evaluated at the Clinic in the year 2000 were revised. This initial assessment is performed by a physician, either an orthopedist or physiatrist, who fills out a specific protocol for the clinic after receiving a specific training at the MMC Service. The selected patients were those who were younger than one year at the first assessment. Any interfering events or error filling out the patients' files were considered exclusion criteria.

RESULTS

A total of 230 patients underwent the initial evaluation at the MMC Clinic of AACD-SP in the year 2000. Of these, 64 (27%) were younger than 1 year of age at this first assessment, with a mean age

of 5 months; 44% of them were females. Two files met the exclusion criteria and were excluded from the study (one patient who had only undergone the triage and another whose file was reissued.

When the origin of these patients was analyzed, we observed that 46% were from the capital city of Sao Paulo (28 patients), 50% were from the countryside of the state of Sao Paulo (31 patients) and 4% were from other states (1 patients from Parana and 2 patients from Minas Gerais).

Regarding the pregnancy information, 13% had been born through normal delivery and (8 cases) and 87% through C-section (54 cases). Prematurity (birth before 37 weeks of gestation) occurred in only 5% of the cases (3 patients). Most parents did not know how to correctly report the Apgar index (fetal distress index). Only 2% of the cases had a previous history of myelomeningocele in a previous pregnancy (1 patient).

Twenty-three patients (37%) did not have a prenatal diagnosis of hydrocephalus and/or myelomeningocele. The diagnosis was attained during pregnancy at the third-trimester ultrasound (US) in 33 % (21 patients), at the second-trimester US in 29% (19 patients) and at the first-trimester US in 1% (01 patient).

When we analyze the age at the closing of the defect, it was observed that in 51% of the patients (32 cases) this occurred within the first 24 hours of life, followed by 32% (20 cases) in the first week, 11% (7 cases) in the second week, 1% (1 case) in the fourth week and only 3% (2 cases) did not need to undergo the closing of the defect, as they had myelodysplasias.

Only 11 patients (17%) had not undergone ventriculoperitoneal shunt (VPS) up to the moment of the initial evaluation. Among the patients with VPS, it was observed that the patients' age when the surgery was performed was 1 to 7 days in 19% (12 patients), 8 to 14 days in 12% (8 patients), 15 to 21 days in 16% (10 patients), 22 to 28 days in 8% (5 patients), 29 to 34 days in 12% (8 patients), 35 to 45 days in 3% (2 patients), 46 to 60 days in 6% (4 patients) and older than 60 days in 3% (2 patients).

When the patients' neurological level at the initial evaluation was analyzed, we found 22 patients with thoracic level (T = 35%), 18 high lumbar (HL = 29%), 15 low lumbar (LL = 24%), 7 asymmetric (A = 11%) and no patient with the sacral level (S). Among the 7 patients with asymmetric level, different combinations of levels were observed on the right and on the left (r/l): 3 patients T/HL, 1 patient HL/LL, 1 patient LL/HL, 1 patient HL/T and 1 patient LL/T.

DISCUSSION

The incidence of myelomeningocele is variable in the literature; in countries such as the United States, it is of 1:1000 live births. In Brazil, there is scant published information on the subject; however, there are two recently published studies, one carried out in Campinas – SP, which shows an incidence of 2.28:1000 births in the period of 1982 to 2001[7] and another in Curitiba – PR, which shows an incidence of 1.8:1000 births in the period of 1990 to 2000.[8]

These data are compatible with those found in Latin America, of 1.5:1000 births.[9] Factors such as lack of access to adequate prenatal care can influence this indicator.[10] Considering that in other countries, where the prenatal diagnosis is attained more often, the parents can choose an elective abortion, the incidence statistics in these countries can be influenced by this factor.

There seems to be a slight predominance of the female sex, at a ratio of 1:1.2[7] to 1:1.5;[11] however, the present study showed the opposite pattern, with a slight predominance of the male sex at a ratio of 1.2:1.

This finding may be due to a bias of this study caused by its small sample size. The chance of recurrence for this malformation within the same family is 5%. However, this sample showed only one case of MMC in a previous gestation. It is known that genetic counseling is fundamental for the parents, a procedure that is routinely carried out at our Service after the initial assessment.

Although the prenatal diagnosis with the morphological US can be performed earlier, from 15-18 weeks of gestation, this study showed an elevated rate of patients who did not have the diagnosis in the prenatal period (37%), or who had the diagnosis only at late pregnancy (33%). A similar aspect was observed in another Brazilian study, where the diagnosis was made, on average, at 29 weeks of gestation.[7] This was an expected finding, as the morphological US assessment with emphasis on fetal morphology is not carried out routinely in Basic Health Units in Brazil, during the prenatal follow-up.

Only 27% of the patients were younger than 1 year old at the initial assessment at the institution, an age during which intervention can more effectively prevent orthopedic deformities, through adequate positioning and orthosis use, in addition to preserving renal function with the use of techniques for the correct vesical emptying. Therefore, it is also possible to follow the evolution of the hydrocephalus and the functioning of the VPS.

In this study 51% of the patients were submitted to early closing of the defect, when they were as young as one day old. It is known that, in some selected cases, an intra-uterus surgery can be performed, but none of our patients underwent intra-uterus repair of the defect. Further studies are necessary to confirm the best surgical technique and the best moment for the surgery to be performed.

The prevalence of obstructive hydrocephalus, caused by the tamponment of the foramen magnum by the cerebellum amygdala, varies in literature from 86%[7], 90%[1] up to 97.5%.[11] We found a similar pattern in the present study, with hydrocephalus being present in 83% of the patients and all had undergone VPS. It is known that the complications caused by the hydrocephalus shunting are important causes of morbi-mortality in the first year of life for these patients, due to infections that cause meningitis and ventriculitis.

The early closing of the defect within the first hours of life and the control of the hydrocephalus with VPS in the first months of life lead to an increase of survival in these patients. Although these treatments are carried out early, the referral to rehabilitation therapy is still slow in our country.

In the world literature, the most prevalent functional neurological levels are at the lumbar (mid-lumbar and low-lumbar) and the sacral levels. A study carried out in Cataluña, Spain, with 322 patients, described that 11.2% were at the thoracic, 10.5% were

at high-lumbar, 23.9% were at mid-lumbar, 36.9% were at low-lumbar and 17.3% were at the sacral level. Even considering the small differences due to the different classification criteria, our study found a higher number of patients at the thoracic (35%) and high thoracic (29%) levels, in relation to the low lumbar (24%) and sacral level (0%).

The interpretation of the authors for this disagreement, when compared to the world literature, is due to the fact that AACD is a reference center in the treatment of MMC and thus, children with higher functional levels are preferably referred to the center, considering that children with lower levels are more functional and the rehabilitation intervention may seem less essential for professionals that are not familiar with the entire array of problems that they can present – deformities, pressure ulcers, loss of renal function, etc. However, other factors must be considered such as, for instance, children with lower levels who might be followed at other Services or who are not adequately diagnosed.

CONCLUSION

This study demonstrates the clinical-functional profile of the patients treated at the MMC Clinic of AACD-SP, which represents only a small number of the Brazilian children with MMC. Further studies are necessary to better understand how these patients are being treated in our country.

Considering that these patients' survival and quality of life are related to the neurological, urological, orthopedic and general complications, it is important to try to stimulate not only the prevention, but also the early diagnosis and treatment of this disease, aiming at decreasing its impact on society. In order to do so, hospitals and maternities, as well as healthcare professionals, should be prepared to suspect and confirm the diagnosis of MMC, referring these patients to a rehabilitation center as soon as possible, regardless of their neurological level.

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