Rare diseases and small maternity units

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SUMMARY: Rare diseases and small maternity units.

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Introduction. Frequent turnover of neo-specialists and high prevalence of obstetric physiology are typical in the maternities of small peripheral hospitals where obstetric and gynaecologic pathologies are prevalently acute. Both implicate an high risk of desuetude in management of important and/or rare pathologies.

Material and method. A case of alobar holoprosencephaly is reported, casually encountered in the prenatal outpatients ambulatory of Asiago Hospital.

Discussion. The rarity of the case generated some awkwardness in the sanitary staff with risk of either under and over-treatment despite the existence of guide-lines and protocols.

Conclusions. Despite the frequent turnover of young specialists in the small peripheral maternity units, we suggest an adequate prenatal echographic training to be a relevant requirement for both early recognition and management of rare pathologies.

KEY WORDS: Holoprosencephaly - Rare diseases - Prenatal early diagnosis.

Oloprosencefalia - Malattie rare - Diagnosi prenatale precoce.

Introduction

The small Maternity Units of small peripheral hospitals play important socio-sanitary roles. These placed in pleasant and touristic areas, as Asiago is, become sites of reference and recovery for obstetric-gynecological tourists which commonly overcome resident popula-

Original article

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435
mary and secondary employment and the emigration of high scholastic level people is prevalent: this implies frequent under-medical staff availability and/or a frequent turn-over of first employ neo-Obstetrics and Gynecology specialists who prefer to move as soon as possible to more glamorous maternity units.

In the small Maternity Units the clinical casuistry is small also: physiology represents the majority being pathology reserved to acute fetal distress during delivery and to acute obstetrics-gynecological abdomens. Regarding to benign, functional and neoplastic surgery the clinical activity is conditioned by the personal experience of single Gynecologists and Anesthetists and by the hospital guide-lines and protocols. All this may imply either a progressive desuetude in the diagnosis and management of important and/or rare pathologies or an under/over treatment in spite of the availability of guide-lines and protocols.

We report a case of alobar holoprosencephaly found in the Obstetrics and Gynecology Unit of Asiago’s Hospital. The absence among the medical and nursing staff of any reminiscence in regard has produced in someone worry, doubts and difficulties in clinical approach and management.

Definition

Holoprosencephaly is a rare pathology (ICD-9-CM 742.2; ICD-10 Q04.2, with synonym Arinephaly Cyclopyn and eponymous Cyclopes) consisting in a complex malformation of the Central Nervous System secondary to an incomplete division of the prosencephalon between 18th and 28th day of gestation (1). Ensue several anomalies implicating various incomplete development of cerebral hemisphere, pellucidum septum, callosus corpus, sagittal scissure, optic and olfactory centers, till hypophysis agenesis with secondary pan-hypo-pituitarism, mono-ventricle encephalon (olosteric) with sinophthalmia (cylopyn) and other face dismorfisms (ipter-telorism, schisis, proboscis, etc.) (2) (Figure 1).

Holoprosencephaly has an incidence between 1/10.000 live/dead born or 1/250 embryos or 1/16.000 (1, 3) to 1/54.000 live born of any sex.

Etiology is heterogeneous: connatal infections (TORCH), generic drugs assumption, maternal diabetes, ionogenic radiations, trisomies and chromosome 18 and 13 deletions, and eponymy Syndromes (Smith-Lemli-Opitz, CHARGe) (2).

The recurrence risk is between 1-6% according to chromosomal or not chromosomal origin of holoprosencephaly.

Case report


The first routine echographic evaluation is performed on august 25, 2014: “Complete thalamus fusion, single ventricle horse-shoe shaped (Figure 2). The assial coronal scanning shows continuity between dorsal sac and mono-ventricle, absence of pellucidum septum, of callosum corpus and of inter-hemispherical scissure (4). The face profile appears flat with single orbit and microcephaly; nuchal translucency mm 3.9 (Figure 3). Crown-ramp length mm 67 (Figure 4). The remaining morphology, placental insert and amniotic fluid appear normal”.

The images show a case of alobar holoprosencephaly horse-shoe shaped and cyclopyn at 12 weeks of gestation.
The case hesitates in a request of voluntary abortion within 90 days (law 194/1978).

Comment

According to the entity of the defect in cerebral hemisphere division, the holoprosencephaly is defined alobar (the worst type, with single ventricle and round skull) and semilobar (with a partial division of posterior hemispheres) both fatal and lobar (with no evidence of the pellucidum septum, difficult to diagnose needs a differential diagnosis with septum-optic dysplasia) consents survival with psychomotor retards and other deficits.

In the worst cases (alobar and semilobar holoprosencephaly) the diagnosis is possible at 9 weeks of gestation (5, 6) by means of Echography and Magnetic Nuclear Resonance (4). Differential diagnosis is with the heavy hydrocephalus, the Walker-Warburg Syndrome, the inter-hemispherical cyst of great dimensions, the otocephaly and other defects of the median line.

The main signs can be cyclopy, proboscis, pre-maxillary agenesis, bilateral and middle labio-palato-schisis, coloboma, retinal dysplasia, choanal stenosis, stenosis of piriform sinus, hypertelorism, single medial incisor tooth, and even a normal face. In case of survival (lobar holoprosencephaly) babies have hydrocephalous, delayed development, motor deficit, oral-motor disorder, epilepsy, hypopituitarism.

Conclusions

With regard to the frequent turnover of neo specialists in small Maternity Units and to the limited pathology casuistry of peripheral small hospitals, we suggest an adequate prenatal echographic training to be a relevant requirement for the recognition and the management of rare pathologies. In fact the early diagnosis of alobar holoprosencephaly permitted to acquire quickly a voluntary abortion (law 194/1978) rather than to refer the patient to higher level maternity unit and to recur to more belated and complex measures.

References