SUMMARY OF THESIS "PLURIMALFORMATIVE SYNDROMES HIGHLIGHTED IN THE NEONATAL PERIOD"

Author: Raluca Marian

Part I of the paper includes summaries of the literature on major development processes of human embryogenesis, plurimalformative syndrome, neonatal and prenatal diagnosis, ethical issues in diagnostic and therapeutic approach to multiple abnormalities.

The first chapter is an introduction to the general prenatal development comprising most fascinating, least understood and most dynamic phase of human development from a single cell and ending with the birth of a child.

In the second chapter we dealt with general aspects of congenital anomalies in terms of epidemiology, risk factors and clinical aspects, following that the third chapter to deal with ethical issues and practical conduct in the diagnosis and prevention of multiple congenital anomalies and plurimalformative syndromes. Delay in diagnosis may delay initiation of therapy where possible, with serious repercussions on disease progression.

In the second part of the paper, after presenting the objectives, the importance of the problem, method of study material and exposed personal research results of the two large studies. Each study group is shown by partial results and conclusions, final conclusions being collated at the end of the thesis. The paper includes an important iconographic material, collected during the four years of study.

The objectives of the study were: to assess the frequency of plurimalformative syndromes and the nonsyndromic critical heart malformations in newborns from a tertiary reference center, highlighting maternal and fetal risk factors, their correlation with the occurrence of this severe pathology, classification of multiple malformation in recognized syndromes, the description of symptoms and evolution of the cases with plurimalformative syndromes for a period of 6 months, estimate the prevalence of known major plurimalformative syndromes: Down syndrome, Patau, Edwards, assessing prevalence and risk factors of nonsyndromic critical congenital heart diseases and diagnostic approach and management of nonsyndromic critical congenital heart diseases.

Material and methods

This study was conducted in the Regional Center of Neonatal Intensive Care Unit (RCNICU) Tirgu Mures in the period 1 January 2007 to 31 December 2010 and it was prospective study. In the Regional Center of Neonatal Intensive Care Unit from Tirgu Mures are cared newborns derived from subordinated counties (Mures, Harghita) and neonates with severe neurosurgical pathology and cardiac malformation in the country. Were followed infants with revealed multiple anomalies, syndromes and critical congenital heart malformations born or transferred to RCNICU Tirgu Mures over this period.

• **The control group** consists of 92 newborns with appropriate adaptation to extrauterine life, with Apgar score at 5 minutes 9-10, derived from healthy mothers or with minor illness during pregnancy. Infants had a favorable outcome during hospitalization (without severe disease) being both mature and premature. They had similar gestational age as the group with plurimalformative syndromes. Thus, we followed 48 mature infants, 44 preterm newborn infants with gestational age (GA) between 27-36 weeks. Term newborns without pathology were cared for rooming-in system, and those with mild illness and premature babies were cared for in a neonatal intensive care department and later rooming-in.

• **Study group I** (group of newborns with plurimalformative syndromes) - patients born or transferred to RCNICU Tg. Mureş, consists of 92 newborns cared in the neonatal intensive care department, where was provided thermal comfort, ventilation and support of an adequate oxygenation, rebalancing electrolyte and acid-base and base pathology specific treatment

• **Study group II** (group of newborns with critical congenital heart diseases not associated with a plurimalformative syndrome) were represented by 86 infants born or transferred to RCNICU Tg. Mures, who were cared for in a neonatal intensive care department.

In the last part of the paper, after analyzing the statistical results of the two studies, were then mapped discussion and final conclusions of this thesis. Some of the most important results and conclusions are:

- The prevalence of congenital anomalies recognized at birth was 62.2 cases per 1000 live births
- Prevalence at birth of plurimalformative syndromes was 9.4 cases per 1000 live births

• *Significant correlations* were found between maternal pathology, consanguinity, the presence of a deformed child in history, mother's rural provenance, chronic intrauterine distress and the frequency of neonate with plurimalformative syndromes. *No significant correlations* were found between maternal age, medication, alcohol, pregnancy follow-up, newborn sex and frequency of plurimalformative syndromes in newborns

• The prevalence of nonsyndromic critical congenital heart malformations presented a significant increase, tripling its value in 2010 (1.42%) compared with 2007 (0.46%). *No significant correlations* were found between maternal age, maternal pathology, pregnancy follow-up, type of birth, sex, birth weight, gestational age of the newborn and the incidence of neonatal plurimalformative syndromes.

• Given the socio-economic and psychosocial impact of congenital anomalies detected at birth, proper evaluation, at country level, of the prevalence at birth of congenital anomalies in general and plurimalformative syndromes in particular, requires a standardized reporting system. It requires reporting of congenital anomalies in a functioning National Register, initially at regional level (pilot studies, multicenter studies), and then at contry level.

• It would be desirable the affiliation of the National Registry of congenital malformations in the European register, EUROCAT

• It is necessary to improve prenatal diagnosis by establishing centers of excellence for perinatology. Last but not least is compulsory an appropriate material support of neonatal intensive care and pediatric cardiovascular centers so that they can cover the necessary treatment in Romania.