DOWN SYNDROME ASSOCIATED WITH THE NEONATE INFECTION PATHOLOGY

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Abstract

The Down syndrome is a chromosomopathy characterized by the presence of a supplementary 21st chromosome, with particular phenotypic appearance.

The authors have an analysis of immediate and from distance prognostic of the patients from the study to which the pathology especially the brunk infection reduces the distance prognostic.

The study was done at the Neonatology and Children Clinics over a period of 4 years. In the study was included 36 patients which presented clinical picture characteristic to the syndrome associated with specific changes of the karyotype.

Newborns with Down syndrome have presented infection pathology associated with malformation syndromes (cardiac) which extend the length of hospitalization on the one hand and clinical-biological recovery on the other hand.

Keywords: Down syndrome, infection, new borns

Introduction

The Down syndrome represents an interdisciplinary complex pathology during childhood period, especially for newborns, with an incidence of 1 out of 700 births(1).

It's a chromosomopathy characterized by the presence of a supplementary 21st chromosome, which, clinically, leads to a complex picture, with particular phenotypic appearance, delay in acquisitions(5, 7) associated or not with isolated or combined cardiac, digestive or osteo - articular malformations,.

For all the women with risk (above 35 year old, existence of trisomy inside the family), the amniocentesis allows prenatal diagnosis (5) and interdisciplinary genetic counseling, but the attitude towards the pregnancy is fully of parents in concordance with moral, religious and ethnic principles of the family.

Objectives

In this paper, the authors have an analysis of the pathology associated with Down syndrome, which leads to an increase of the hospitalization period and complications. In the same time it is realized an analysis of immediate and from distance prognostic of the patients from the study group to which the pathology especially the bunk infection reduces the distance prognostic.

Materials and methods

The study was done at the Neonatology and Children Clinics over a period of 4 years. From a total of 3270 patients, 36 were included in the study which presented clinical picture characteristic to the syndrome associated with specific changes of the karyotype (fig. 1).

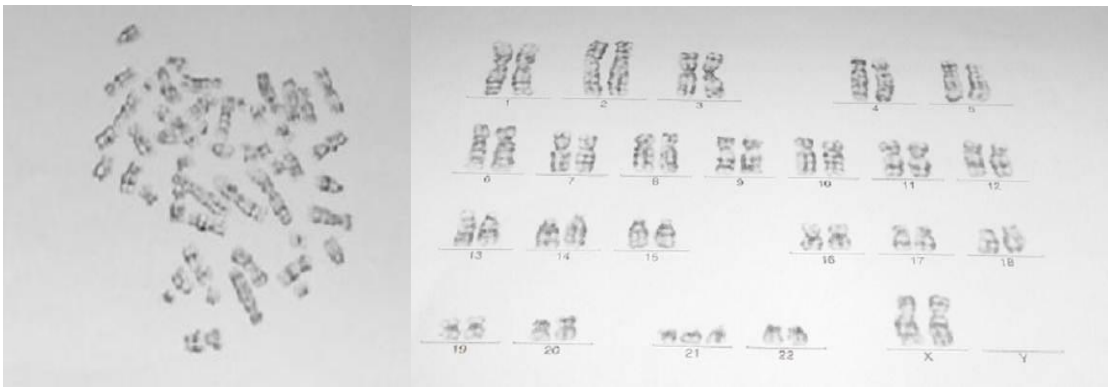

Fig. 1. Karyotype 47, xx+21.

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Cytogenetic diagnostic of 21 trisomy was presents in 34 children (94.44%) and only 2 cases (5.56%) had the mosaic. The investigations were done precocious postnatal after the specific clinical signs were indentified.

The work method was represented by retrospective analysis of patients' observations papers and also by dynamic analysis of clinical and biological picture of hospitalized patients (6 cases) from the beginning of the study.

In the study were included patients with: typical malformation phenotype, specific changes of the karyotype, clinical and biological picture which complements the positive diagnosis.

Complex laboratory investigations and multiple interdisciplinary consults were done: genetic, cardiologic, endocrinology, pediatric surgery.

Results and discussions

The presence of homogenous trisomy 21 was high to the group of study (94.44%) similar with the data from literature (90-95%) (6), unlike the mosaic (5.56%) (fig. 2) higher than literature (1-2.5%) (7). Inside the group of study we didn't have cases of translocation.

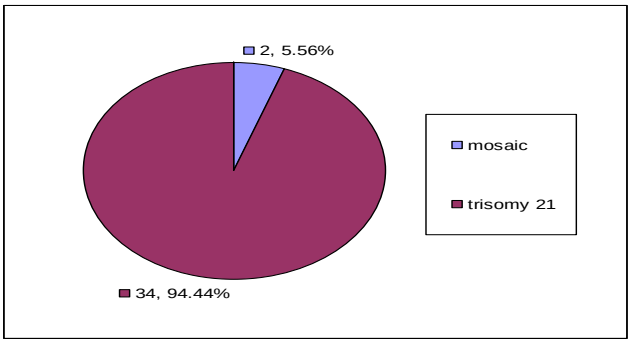

Fig. 2. Cases repartition after karyotype aspect.

The analyses of personal history, for placement in categories of newborns highlights a high proportion of premature children, 22 cases with gestational age less than 37 weeks and 14 term infants. Regarding the birth weight in 77.77% of cases the intrauterine growth retardation was present.

The phenotypic appearance is characterized by: brachycephalic head, with flattened occiput and broad fountains (7); round face "moon face", flat (7); epic (1); oblique fissure vents up and out (1); small nose with flattened root , small nostrils and anteversion (5) was present all the cases from the study.

Some clinical signs like open mouth, with tongue protrusion – pseudomacroglosie (6); small, dysplastic and lower inserted ears (7); short neck, with excess skin on the scruff (6); short, broaded and with brachydactyly hands (shortened fingers), clinodactyly (no bowing fifth finger) and a single palmar flexion crease (simian crease) (7) were present in varying proportions, except the simian fold, frequent sign to the patients with Down malformation, present in 78% of cases, which coincides with literature numbers.

Mental retardation present in all patients ranged from severe to moderate.

The pathology malformation was present, single or in combination, the proportion of patients without any malformation was little (6.7%), higher than the literature data (5.9%) (2), remark for predominance of cardiac malformations (ventricular septal defect - 44.44% vs.12-35% (2), atrial septal defect - 19.44% vs.5-38% (2), atrio-ventricular canal - 16.66% vs.3-55% (2)), musculoscheletal malformations (congenital clubfoot – 30.55% , polydactyly - 8.66%, syndactyly- 5.55%) (fig. 3).

The gastrointestinal anomalies were present in the proportion of 13.86% vs. 4-11% (2) to five patients (two patients with duodenal stenosis surgery, a patient transferred in Pediatric Surgery Service, a case of duodenal atresia type one surgery, a case of paralytic ileus).

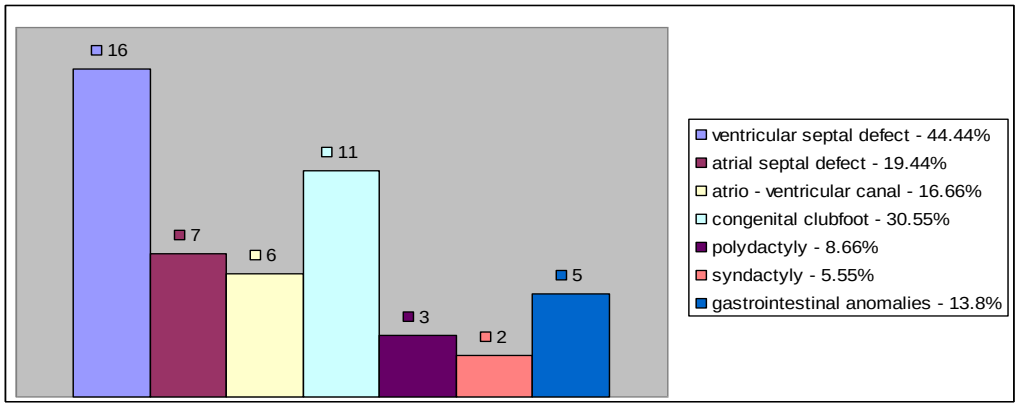

Fig. 3. Cases repartition depending on associated malformation.

All patients under study had deficient diet, absent sucking reflex – 72.22% cases, sucking - swallowing incoordination, that determinates a constant weight deficit, with growth disorders, weight curves slowly ascending, which leads to increased length of hospitalization.

Average length of stay to children with Down malformation was 45-60 days, in contrast to literature data which states much lower average length about a week (5) to the newborns at term with Down Syndrome.

The increased of average length of stay in association with immunological deficits specific to neonatal and child at breast period on the one hand, and the other hand associated to the affection, have increased the incidence of infections and the growing of their severity.

The specific immunodeficiency to Down syndrome includes following association: mild to moderate lymphopenia of T and B cells, with marked decrease of naïve lymphocytes, the affect of proliferation of T cell mitogen-induced, the reduction of specific antibody responses to vaccinations and defects of neutropfil chemotaxis. Secondary immunodeficiency due to nutritional or metabolic factors in Down syndrome and the deficiency in zinc were cited. Non-immunological factors, including abnormal anatomic structures (for example the small ear canal, tracheomalacia) and the gastro-esophageal reflux play a role in increasing the frequency of respiratory tract infections, found in our study for 2 cases. The molecular mechanism leads to immune defects observed to individuals with Down syndrome and the contribution of these immunological anomalies to the increased risk of infections requires further investigation. The approach of immunological and non-immunological factors involved in the pathology of the infectious diseases can reduce the infections susceptibility to individuals with Down syndrome (4).

From the group of study 3 cases had an unfavorable evolution to death due to pathology of associated malformation: cord malformation, neonatal sepsis and necrotic enterocolitis ulcerative.

So, to the group of study the neonatal septicemia was present at 36.11% cases, whereas localized lighter forms were: congenital pneumonia – 16.66%, necrotic - ulcerative enterocolitis -11.11%, other infections 36.1% (fig. 4).

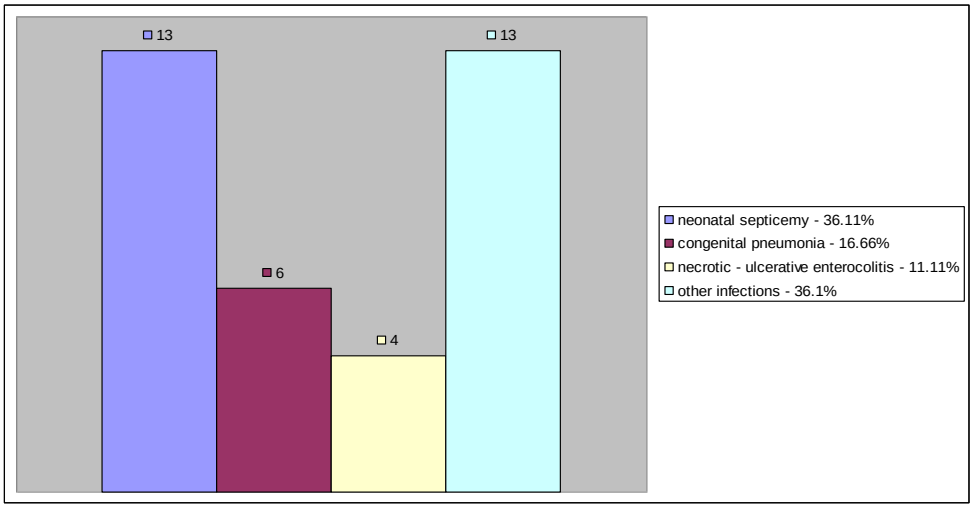

Fig. 4. Infection cases repartition.

The septicemia cases have presented train evolution which resulted in clinical and biological late recovery with increasing duration and cost of hospitalization.

Diagnosis was easily established based on positive cultures, positive blood cultures, evidence of positive rapid inflammatory (persistent and severe thrombocytopenia, highly positive C-reactive protein, positive procalcitonin with values over 7-8-12 ng/ml, severe leukocytosis).

From bacteriological point of view in neonatal septicemia with early onset the results attest presence of gram-negative bacilli in proportion of 94% with predominance of Pseudomonas Aeruginosa (44%), followed by Serratia Marcenses and Klebsiella Pneumoniae and in 6% of cases the coagulase-negative staphylococcus is present (fig. 5). According to the specific literature, in 30-40% cases the principal factor involved in neonatal septicemia with early onset is the group B streptococcal (4).

In septicemia with late onset germs most frequently involved are also gram-negative bacilli (77%), but the most common is Serratia Marcenses, followed by Klebsiella Pneumoniae and Pseudomonas Aeruginosa (fig. 6). 23% from microbial flora is formed by gram-positive cocci. (3)

Regarding the onset: 5 cases have presented early onset in the first 5 days of life, what sustains intrauterine infection of the newborn, 8 cases have presented late onset due to prolonged hospitalization, specific immunodeficiency and associated malformation syndromes.

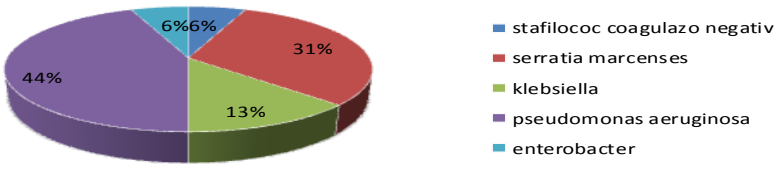

Fig. 5. Incidence of microbial flora in early onset septicemia.

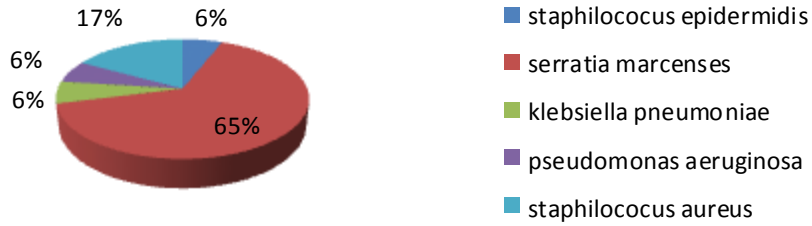

Fig. 6. Incidence of microbial flora in late-onset septicemia.

Conclusions

1. Newborns with Down syndrome have presented infection pathology associated with malformation syndromes (cardiac) which extend the length of hospitalization on the one hand and clinical - biological recovery on the other hand.

2. Cytogenetic diagnosis made earlier and to all patients from study highlights a predominance of trisomy 21 (94.44%) and the mosaic (5.56%). We don't have translocation cases to patients from the study.

3. For group of study we didn't observed a correlation between the intensity of phenotypic characters, the prevalence and the intensity of clinical and biological manifestations. But it was observed a correlation with the associated malformations, their intensity, evolution and prognostic beeing much hampered to patients with associated malformations.

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