

The prevalence and nosological structures of primary immunodeficiencies among children in the Altai region

SCO — краткое сообщение

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Распространенность и нозологическая структура первичных иммунодефицитных состояний среди детей Алтайского края

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Primary immunodeficiencies (PIDs) are a heterogeneous group of congenital diseases of the immune system, associated with the loss, reduction or malfunction of one or several its elements and more than 450 current nosologies. For a long time PIDs have been classified as orphan diseases. However, with the introduction of neonatal screening in a number of countries it has become obvious that PIDs prevalence is not so rare. According to the European Register, created with the support of the European Society for Immunodeficiencies (ESID) with its 28 European States [1], PIDs prevalence is 3 cases per 100,000 people in Germany [2], 6 cases per 100,000 people in Switzerland [3], 7 cases per 100,000 people in France [4]. In Russia the National PID Patient Register was launched on the “Medical online platform ROSMED.INFO” in 2019, established under the auspices of the National Expert Association of Primary Immunodeficiencies. According to the Register, PIDs prevalence in Russia varies significantly by region (from 0,9 to 2,8 per 100,000 people) and is on average $2,09 \pm 0,93$ per 100,000 people [5, 6]. The annual PIDs incidence in Russia is $5,7 \pm 0,6$ per 100 live births. The article presents the analysis of PIDs prevalence and structure among children in the Altai region.

Objective — to study prevalence and nosological structure of PIDs among children in the Altai region.

Materials and methods. The information about children with PIDs is taken from the PIDs Patient Register on “the Medical online platform ROSMED.INFO”. The data about children with PIDs, living in the Altai region, have been collected since 2019 till now. An obligatory condition of including patients in the Register was providing informed content by a legal representative, according to the Russian law № 152-FL on personal data protection. The Register included all children with diagnosed and genetically unverified PIDs at the time of registration.

Results

Epidemiology. PIDs Patient Register includes 23 children, living in the Altai region. PIDs prevalence among the child population of Altai region is 4,8% cases per 100,000 children (the child population in the Altai region for 2022 is 482,000).

Structure. The analysis of PID nosological structure showed that in the region there are more cases of PIDs with immune dysregulation — 5 (21,7%) and autoinflammatory diseases — 5 (21,7%), the second place by frequency of combined PIDs with syndromic pathology — 4 (17,4%) and PIDs with

a dysfunction of antibody formation — 4 (17,4%), PIDs with the complement system defect account for 2 (8,7%) cases, unspecified PIDs in 3 (13,0%) children. 18 (78,3%) patients passed molecular genetic analysis.

Demographics. Out of 23 children with PIDs, registered in the regional Register, 22 (95,6%) live, 1 (4,4%) dead. The age structure of children with PIDs: 6 (26,1%) children aged 0-5 years, 9 (39,1%) children aged 6-10 years, 8 (34,8%) children aged 11-18. The youngest patient at the time of analysis was 1 year old. Out of 23 patients 20 (86,9%) male children and 3 (13,1%) female children. Most children with PIDs are residents of large cities in the Altai region — 14 (60,8%), 9 (31,2%) patients live in rural areas.

Delay in diagnosis. In most cases PIDs were registered in the first 5 years of life — 9 (39,1%) cases, up to 1 year — 3 (13,0%) cases, 1-4 years — 6 (26,1%) cases, 5-10 years — 8 (34,7%) cases, 11-18 years — 6 (26,1%) cases. More frequent cases of combined PIDs are diagnosed in the early stages. Most patients had significant delay in diagnosing PIDs compared to the first clinical manifestations of the disease and varied from 1 months to 4 years.

Therapy. A regular substitution therapy with immunoglobulin products is required to 10 (43,5%)

children, 9 of which get subcutaneous immunoglobulin (SCIG), 1 — intravenous (IVIG). The children receive immunoglobulin products regularly. 4 (17,4%) children receive chemotherapy, 1 (4,3%) child — anticytokine therapy, 1 (4,3%) patient with inherited angioedema gets C1-inhibitor and an inhibitor of a bradykinin receptor during attacks.

Conclusion. PIDs prevalence among the children in the Altai region was 4,8% cases per 100,000 of child population that is above the average PIDs prevalence in Russia — 2,09 per 100,000 people [6]. PIDs structure among the children in the Altai region differs from PIDs structure according to the National Registry. While PIDs with a dysfunction of antibody formation and combined with syndromic pathology prevail in the National Registry, PIDs with immune dysregulation and autoinflammatory diseases prevail in children with PIDs, living in the territory of the Altai region, and PIDs with a dysfunction of antibody formation and syndromic pathology take the second place in frequency. Regular replacement therapy with immunoglobulin is given to all children in need of this treatment. Most of them 9 (90%) get SCIG and 1 (10%) child — IVIG. The treatment is received regularly. 78,3% of patients obtained molecular genetic examination..

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