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INCIDENCE OF SLAVIC MUTATION IN NIJMEGEN BREAKAGE SYNDROME GENE IN EAST SLOVAKIAN POPULATION

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Nijmegen Breakage syndrome (NBS) is one of disorders characterized by radiosensitivity, immunodeficiency and higher occurrence of chromosomal breaks with high risk of malignancy, mainly of lymphoreticular system. NBS gene is located on chromosomel region 8q21 and is encoding nibrin - a protein playing an important role in DNA breaks repair.
The incidence of NBS is estimated to be 1:100 000 - 1:270 000; most of the patients originate from the middle and east European regions inhabited by Slavic ethnic and have a "Slavic" mutation - 657-661delACAAA. The incidence of heterozygots for the Slavic mutation is estimated to be 1:177, in the East European region (Ukraine, Poland, Belorussia) even more - 1:80. However, until the beginning of our study there was no diagnosed patient with NBS in the East Slovakia. Therefore we intended to ascertain whether there is a really low incidence of the NBS gene or just an underestimated diagnostics.
We examined the population of all neonates born from February 2001 to January 2002 in Faculty Hospital in Košice. The obtained results were surprising - in the whole file there was no sample positive for the Slavic mutation. We came to the conclusion that East Slovakia does not belong to the Slavic regions with high incidence of the mentioned mutation". Despite this fact the importance of diagnosing NBS should be kept in mind since the early management could lower the risk of malignant complications in these patients.