# ПУБЛИКАЦИИ ОСНОВНЫХ РЕЗУЛЬТАТОВ НАУЧНОЙ ДЕЯТЕЛЬНОСТИ СОТРУДНИКОВ МЕДИЦИНСКОГО ИНСТИТУТА В РЕЙТИНГОВЫХ ЖУРНАЛАХ, ИНДЕКСИРУЕМЫХ В БАЗАХ ДАННЫХ WEB OF SCIENCE / SCOPUS ЗА ПЕРИОД С 2017 ПО 2020 гг.

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1. Kondo, H, Maksimova, N, Otomo, T, Kato, H, Imai, A, Asano, Y, Kobayashi, K, Nojima, S, Nakaya, A, Hamada, Y, и др. Mutation in VPS33A affects metabolism of glycosaminoglycans: a newtype of mucopolysaccharidosis with severe systemic symptoms // HUMAN MOLECULAR GENETICS. – 2017. – Vol. 26. – Iss. 1. DOI: 10.1093/hmg/ddw377. База данных: Scopus/Web of Science. Квартиль: Q1.

Аннотация:

Mucopolysaccharidoses (MPS) are a group of genetic deficiencies of lysosomal enzymes that catabolize glycosaminoglycans (GAG). Here we describe a novel MPS-like disease caused by a specific mutation in the VPS33A gene. We identified several Yakut patients showing typical manifestations of MPS: coarse facial features, skeletal abnormalities, hepatosplenomegaly, respiratory problems, mental retardation, and excess secretion of urinary GAG. However, these patients could not be diagnosed enzymatically as MPS. They showed extremely high levels of plasma heparan sulphate (HS, one of GAG); 60 times the normal reference range and 6 times that of MPS patients. Additionally, most patients developed heart, kidney, and hematopoietic disorders, which are not typical symptoms for conventional MPS, leading to a fatal outcome between 1 and 2-years old. Using whole exome and Sanger sequencing, we identified homozygous c.1492C>T (p.Arg498Trp) mutations in the VPS33A gene of 13 patients. VPS33A is involved in endocytic and autophagic pathways, but the identified mutation did not affect either of these pathways. Lysosomal over-acidification and HS accumulation were detected in patient-derived and VPS33A-depleted cells, suggesting a novel role of this gene in lysosomal functions. We hence propose a new type of MPS that is not caused by an enzymatic deficiency.

2. Sivtseva, T.M., Vladimirtsev, V.A., Nikitina, R.S., Davidova, T.K., Popov, D.A., Osakovsky, V.L. Intrathecal synthesis of oligoclonal IgG in patients with Viliuiskencephalomyelitis: The relationship between oligoclonal bands and clinical features // JOURNAL OF THE NEUROLOGICAL SCIENCES. – 2018. – Vol. 384. DOI: 10.1016/j.jns.2017.11.030. База данных: Scopus/Web of Science. Квартиль: Q1-Q2.

Аннотация:

Viliuisk encephalomyelitis (VE) is a neurodegenerative disease that afflicts aboriginal people of Yakutia in Siberia with unknown etiology. Oligoclonal IgG bands (OCBs) were discovered in the VE patients (Green et al., 2003). In this study we analysed the association of OCBs with clinical symptoms in 58 VE patients. Positive oligoclonal IgG are associated with a shorter duration of disease (p = 0.002), older age of onset (p = 0.023) and high frequency of main neurological VE symptoms such as dementia, frontal dysbasia, bulbar disorders, muscle atrophy and centrally caused pelvic disorders. Our results show that the OCBs in VE patients are associated with more severe central nervous system (CNS) damage and may cause secondary complications in the course of the disease.

3. Levy, SB, Klimova, TM, Zakharova, RN, Federov, AI, Fedorova, VI, Baltakhinova, ME, Leonard, WR. Brown adipose tissue, energy expenditure, and biomarkers of cardio-metabolic health among the Yakut (Sakha) of northeastern Siberia // AMERICAN JOURNAL OF HUMAN BIOLOGY. – 2018. Vol. 30. – Iss. 6. DOI: 10.1002/ajhb.23175. База данных: Scopus/Web of Science. Квартиль: Q1- Q3.

Аннотация:

Objectives This study provides the first investigation of non-shivering thermogenesis (NST) and brown adipose tissue (BAT) activity among an indigenous circumpolar population, the Yakut of northeastern Siberia. The study also examines the health significance of BAT activity in this population by testing the relationships between BAT thermogenesis and biomarkers of cardio-metabolic disease risk, such as percent body fat and blood glucose and cholesterol levels. Methods Results Data were collected in the Sakha Republic (Yakutia) for 31 men and 43 women. Change in energy expenditure and BAT thermogenesis were quantified after a 30-minute mild cooling condition. Anthropometric dimensions, blood glucose, and lipid levels were also collected. On average, the skin temperature of the supraclavicular area was constant after cooling while the skin temperature of a point on the sternum dropped significantly (P < .001), thus suggesting the presence of active supraclavicular BAT among Yakut adults. Participants with evidence of greater BAT thermogenesis exhibited a larger percent change in energy expenditure (% Delta EE) and an increase in respiratory quotient (RQ) after cooling (P <= .05). While there was no relationship between BAT activity and blood lipid levels, BAT thermogenesis was positively associated with blood glucose levels (P < .01). Conclusions Yakut adults exhibit evidence of active BAT deposits. Given that there is a significant relationship between BAT activity and % Delta EE, it is possible that BAT plays a role in NST among Yakut adults. While the relationship between BAT and body composition is inconclusive, participants with greater BAT seemed to preferentially utilize glucose during cold stress exposure.

4. Zharmagambetova, A, Tuleutayeva, S, Akhmetova, S, Sumanova, A, Baigulakov, A, Sakenov, T, Gorbatova, MA, Grjibovski, AM. Prevalence and experience of dental caries among 12-and 15-year-oldadolescents in Central Kazakhstan // PUBLIC HEALTH. – 2017. Vol. 151. DOI: 10.1016/j.puhe.2017.06.021. База данных: Scopus/Web of Science. Квартиль: Q2-Q3.