

# Trendy vo výskyte Creutzfeldtovej-Jakobovej choroby na Slovensku v rokoch 2007–2022

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Creutzfeldtova-Jakobova choroba (CJch) je rýchlo progredujúce neliečiteľné neurodegeneratívne ochorenie, ktoré patrí do skupiny prionových chorôb. Predkladaná práca sa zameriava na výskyt jednotlivých foriem CJch na Slovensku a analyzuje trendy ochorenia v priebehu rokov 2007 – 2022. V danom období bolo vyšetrených spolu 6 685 vzoriek (3 127 vzoriek krvi, 3 155 vzoriek likvoru, 403 vzoriek mrazeného aj fixovaného mozgového tkaniva). Diagnóza CJch bola potvrdená v 287 prípadoch. Genetická forma ochorenia (gCJch) s mutáciou E200K na prionovom géne tvorila 74 % všetkých prípadov, sporadická forma CJch (sCJch) sa vyskytla v 26 %. Incidencia ochorenia má pri oboch formách CJch stúpajúci trend, v porovnaní s predchádzajúcimi časovými úsekmi je nárast incidence štatisticky výrazne signifikantný ( $p > 0,001$ ). Práca dokumentuje aj nárast veku postihnutých osôb, pri sCJch je priemerný vek 65,3 roka, v prípade gCJch (61,9 roka) je vekový nárast štatisticky signifikantný ( $p = 0,003$ ). Analýza výskytu ochorenia z geografického hľadiska signalizuje pretrvávajúci zvýšený výskyt gCJch v Žilinskom a Banskobystrickom kraji, sCJch sa v zvýšenej miere vyskytuje v Bratislavskom, Trenčianskom a Banskobystrickom kraji.

**Kľúčové slová:** prionové choroby, Creutzfeldtova-Jakobova choroba, mutácia E200K, polymorfizmus M129V, incidencia CJch.

## Trends in the occurrence of Creutzfeldt-Jakob disease in Slovakia in the years 2007–2022

Creutzfeldt-Jakob disease (CJD) is rapidly progressive, incurable neurodegenerative disorder which belongs to the group of prion diseases. The presented study focuses on the occurrence of each form of CJD in Slovakia and analyses its trends during the years 2007–2022. A total of 6685 samples (3127 blood samples, 3155 cerebrospinal fluid samples, 403 samples of frozen and fixed brain tissue) were examined in the given period. The final diagnosis of CJD was confirmed in 287 cases. The genetic form of the disease with E200K mutation on the prion gene was encountered in 74 % of all cases, 26 % was the sporadic form of CJD. Incidence of the disease has an increasing trend in both forms of CJD, compared to earlier periods. The increase in incidence is statistically significant ( $p > 0.001$ ). This study also documents the increase in age of diseased persons, the average age at onset sCJD was 65.3 years, in the case of gCJD the age increase is statistically significant (61.9 years,  $p = 0.003$ ). Analysis of the occurrence of the disease from geographical point of view shows constant increased occurrence of gCJD in Žilina and Banská Bystrica regions. Sporadic CJD has increased occurrence in Bratislava, Trenčín and Banská Bystrica regions.

**Key words:** Prion diseases, Creutzfeldt-Jakob disease, E200K mutation, M129V polymorphism, CJD incidence.

### DECLARATIONS:

#### Declaration of originality:

The manuscript is original and has not been published or submitted elsewhere.

#### Ethics approval and consent to participate:

The authors attest that their study is in compliance with human studies committees and animal welfare regulations of the authors' institutions as well as with the Food and Drug Administration guidelines, including patient consent where appropriate. The authors also declare that their paper is in accordance with the World Medical Association Declaration of Helsinki on Ethical Principles for Medical Research Involving Human Subjects adopted by the 18th WMA General Assembly in Helsinki, Finland, in June 1964, with subsequent amendments, as well as with the ICMJE Recommendations for the Conduct, Reporting, Editing, and Publication of Scholarly Work in Medical Journals, updated in December 2018.

#### Conflict of interest:

Not applicable.

#### Consent for publication:

Not applicable.

#### Authors' contributions:

DZ – supervised the study, analysed the data and wrote the manuscript; MS and AJ – analysed the data; SK, KM and ZS – performed analyses; All authors discussed the results and contributed to the final manuscript.

Cit. zkr: *Neurol. praxi.* 2024;25(1):15-18  
<https://doi.org/10.36290/neu.2023.078>

Článek přijat redakcí: 26. 9. 2023

Článek přijat k publikaci: 4. 12. 2023

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