Genetika Alzheimerovej choroby a demencie s Lewyho telieskami

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Genetika neurodegeneratívnych demencií je turbulentnou témou súčasnosti. Na jednej strane sa postupne zvyšuje množstvo génov zapojených do patogenézy neurodegeneratívnych procesov, na druhej strane vystupuje problém s interpretáciou dosiahnutých výsledkov. Alzheimerova choroba (AD) a demencia s Lewyho telieskami (DLB) predstavujú v súčasnosti dobre definované klinické jednotky. Alzheimerova choroba má jasne určené kauzálne gény (*APP, PSEN1, PSEN2*) a významný gén susceptibility (*APOE*). Popri nich sa postupne objavujú nové gény susceptibility, ktoré modifikujú klinický obraz, vek nástupu ochorenia a spolu s *APOE* vytvárajú komplikované genetické pozadie. Demencia s Lewyho telieskami je heterogénnejšou entitou ako Alzheimerova choroba z klinického aj genetického hľadiska. Génmi susceptibility DLB sú viaceré gény zdieľané s Alzheimerovou chorobou, Parkinsonovou chorobou (PD), frontotemporálnou demenciou (FTD) a inými neurodegeneráciami. V našom príspevku sa snažíme sumarizovať genetické pozadie AD a DLB, charakterizovať ich podobnosti a rozdiely a poukázať na komplexnosť neurodegeneratívneho ekosystému („*neurodegeneratómu*“).

Kľúčové slová: Alzheimerova choroba, apolipoproteín E, demencia s Lewyho telieskami.

The genetics of Alzheimer's disease and dementia with Lewy bodies

The genetics of neurodegenerative dementias is a turbulent topic. On the one hand, the number of genes involved in the pathogenesis of neurodegenerative processes is gradually increasing, on the other hand, the problem of interpretation of the results is emerging. Alzheimer's disease (AD) and dementia with Lewy bodies (DLB) represent currently well-defined clinical entities. AD has clearly defined causal genes (*APP, PSEN1, PSEN2*) and a major susceptibility gene (*APOE*). In addition to these, new susceptibility genes are gradually emerging that modify the clinical picture, the age of onset and, together with *APOE*, create a complicated genetic background. Dementia with Lewy bodies (DLB) is a more heterogeneous entity than Alzheimer's disease, both clinically and genetically. DLB susceptibility genes are multiple genes shared with Alzheimer`s disease, Parkinson disease, frontotemporal dementia (FTD) and other neurodegenerations. In our paper, we aim to summarize the genetic background of both AD and DLB, to characterize their similarities and differences, and to highlight the complexity of the neurodegenerative ecosystem („*neurodegeneratome*“).

Key words: Alzheimer's disease, apolipoprotein E, dementia with Lewy bodies.

Úvod

Alzheimerova choroba a demencia s Lewyho telieskami sú dve najčastejšie neurodegeneratívne demencie. Alzheimerova choroba predstavuje približne 50 – 60 % všetkých demencií a demencia s Lewyho telieska-

DECLARATIONS:

Declaration of originality:
The manuscript is original and has not been published or submitted elsewhere.

Ethical principles compliance:
The authors attest that their study was approved by the local Ethical Committee and is in compliance with human studies and animal welfare regulations of the authors' institutions as well as 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, including patient consent where appropriate.

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