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Cadasil – an Updated Revision

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Background

Cerebral autosomal dominant arteriopathy with subcortical infarct and leukoencephalopathy (CADASIL) is the most common hereditary subcortical vascular dementia, and it is caused by a broad spectrum of mutations in the NOTCH3 gene on chromosome 19.

Aims

The authors intend to review the clinical patterns of the condition and expose the variability of its radiological and genetic data. Furthermore, its diagnostic strategies and available treatments are also clarified.

Methods

Brief description of a clinical case and review of literature collected from online medical databases under the keywords 'CADASIL', 'dementia” and 'genetic”.

Results (including clinical vignette)

The authors report a case of a 67-year-old woman with history of late onset migraine and polymorphic psychiatric and neurologic symptoms, which further evaluation confirmed the CADASIL diagnosis. The involutive course of her condition is also emphasized as well as its devastating social and familiar consequences.
Literature on CADASIL is vast and confirm that its phenotype is variable between and within affected families, with new clinic features continually arising. Nevertheless, main pattern consists in migraine with aura (often atypical or isolated), strokes, cognitive decline/dementia and psychiatric symptoms. Diagnosis is confirmed either by identifying a pathogenic NOTCH3 mutation or by the demonstration of specific granular osmiophilic material in skin biopsies. So far, only symptomatic treatment is available.

Conclusion

CADASIL is notably underrecognized and underdiagnosed. Authors consider it deserves greater awareness from psychiatrists, since it brings great suffering for the patients and their families, as well as high demand for non-psychiatric care.

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