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Images of Interest / Imagens de Interesse

Autosomal Dominant Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) Associated with Multiple System Cerebellar Atrophy (MSA)

Arteriopatia Cerebral Autossómica Dominante com Enfartes Subcorticais e Leucoencefalopatia (CADASIL) Associada a Atrofia Cerebelosa de Múltiplos Sistemas (MSA)

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Abstract

CADASIL, an autosomal dominant genetic disease linked to NOTCH3 gene mutations, manifests itself with small vessel brain infarctions. MSA is a group of neurodegenerative syndromes characterized by autonomic dysfunctions, cerebellar abnormalities, parkinsonism, and corticospinal degeneration.
A 66-year-old woman presented with episodes of altered balance and gait after cerebral ischemia, maintaining episodes of imbalance and lipothymia; ataxic gait, dysmetria and extrapyramidal signs. Prior family history was positive for ischemic brain disease. Genetic study identified mutation in the NOTCH3 gene, in MRI, signs of periventricular microangiopathy compatible with CADASIL and signal alteration in the pons, in the cross aspect, were identified.

Keywords

CADASIL; AMS; Images.

Resumo

Arteriopatia Cerebral Autossómica Dominante com Enfartes Subcorticais e Leucoencefalopatia (CADASIL) é uma doença genética autossómica dominante ligada a mutações no gene NOTCH3, se manifesta com enfartes cerebrais de pequenos vasos. Atrofia Cerebelosa de Múltiplos Sistemas (MSA) é um grupo de síndromes neurodegenerativas caracterizadas por disfunções autonómicas, anomalias cerebelosas, parkinsonismo e degeneração corticoespinhal.
O caso relata uma mulher de 66 anos, que apresentou episódios de alteração do equilíbrio e da marcha, após isquemia cerebral, mantendo episódios de desequilíbrio e lipotimia; marcha atáxica, dismetria e sinais extrapiramidais. Havia antecedentes familiares de doença isquémica do cérebro. Um estudo genético identificou a mutação no gene NOTCH3, na ressonância magnética, foram identificados sinais de microangiopatia periventricular compatíveis com CADASIL e alteração de sinal na ponte, no aspecto cruzado.

Palavras-chave

CADASIL; AMS; Imagens.

Autosomal Dominant Cerebral Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) is an autosomal dominant inherited disease linked to mutations of the NOTCH3 gene that manifests with small vessel cerebral infarctions.[1-3] Multiple System Cerebellar Atrophy (MSA) is a group of neurodegenerative syndromes characterized by autonomic dysfunctions, cerebellar anomalies, parkinsonism and spinal cortical degeneration.[2]

We describe a case of coexistence of MSA and CADASIL that presents clinical and radiological similarities with the only report described in the literature.[2] Here, ischemic events, stiffness, hypokinesia, dysarthria, hyperreflexia, bradykinesia and bradypsychism were detected. This case presents the neuroradiological changes described (Figure 1) and the genetic analysis with identification of the NOTCH 3 gene mutation that supported the high clinical suspicion of CADASIL.

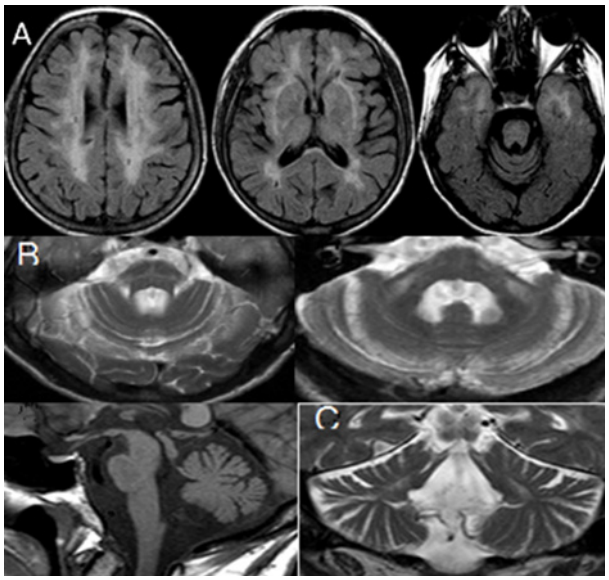

Figure 1 – Skull MRI. A: cerebral volumetric reduction, extensive hypersignal area in periventricular white matter, frontoparietal subcortical; hypersignal in T2, with a cruciform aspect in the pons. B: Findings related to MSA with a cruciform-looking sign anomaly on the pons indicating degeneration of the transverse fibers and median raphe. Signal anomaly and atrophy of the middle cerebellar peduncles and moderate atrophy of the pons. C: Moderate atrophy of the cerebellar hemispheres, with widening of the grooves and compensatory dilation of the IV ventricle.

Ethical disclosures / Divulgações Éticas

Conflicts of interest: The authors have no conflicts of interest to declare.

Conflitos de interesse: Os autores declaram não possuir conflitos de interesse.

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Confidentiality of data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

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