Case 16485

Eurorad

CADASIL: A mimicker of multiple sclerosis

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Section: Neuroradiology
Area of Interest: Head and neck Neuroradiology brain
Procedure: Diagnostic procedure
Imaging Technique: MR
Special Focus: Genetic defects Ischaemia / Infarction
Case Type: Clinical Cases
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Patient: 30 years, male

Clinical History:

A 30-year-old man presented with dysphasia and psychomotor slowing. There was no preceding cognitive decline. The patient was known with migraine with prolonged aura and there was a family history (mother and maternal uncle) of migraine. There were no vascular risk factors. Earlier genetic work-up revealed a NOTCH3-mutation.

Imaging Findings:

CT angiography of supra-aortic vessels was unremarkable. CT of the brain showed multiple bilateral hypodense foci in the white matter of the cerebrum, most of them in supraventricular regions. MRI reveals multiple recent small ischaemic lesions in the semioval centre and corona radiata bilaterally, mostly in the frontal lobes (Fig. 1 and 2). There are confluent white matter alterations in the anterior regions of both temporal lobes (Fig. 3). In addition, there are multiple subcortical -and white matter lesions in keeping with old ischaemia (Fig. 2) and some microbleeds in the basal ganglia and right thalamus (Fig. 4). No significant changes are present in the insular regions and external capsules. There are only limited alterations in the corpus callosum. There are no changes in the U-fibers and the infratentorial white matter is also spared.

Discussion:

CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy) is an autosomal dominant genetic disease of small and medium sized cerebral arteries, and represents the most common genetic cause of ischaemic stroke [1,2]. The estimated minimum prevalence is between 2 and 5 / 100 000 [1]. A mutation in the NOTCH3 receptor gene leads to deposition of granular osmiophilic material (GOM) in the media of the arteries, causing degeneration of vascular smooth muscle cells with mural fibrosis, stenosis and loss of autoregulation, resulting in ischaemia [1-4]. The deposition of GOM is also present in other tissues, but almost always without clinical manifestations [1,2]. Typical clinical manifestations include migraine with aura – often first manifestation, recurrent ischaemic strokes due to lacunar infarcts, mood disorders, progressive cognitive impairment and sometimes acute encephalopathy and seizures [1-3]. There is often a positive familial clinical history of migraine, stroke and dementia [1,2]. Immobility, infections and a build-up of morbidities lead to premature death [2,3]. Imaging features include bilateral white matter abnormalities, lacunar infarcts and sometimes microbleeds [1-

3]. White matter abnormalities consist of multifocal bilateral, symmetrical hyperintensities on T2- Weighted Images (WI) and Fluid Attenuated Inversion Recovery (FLAIR), mostly located in the periventricular and deep white matter [1-3]. There is a predilection for the external capsules and the anterior temporal lobes, with the highest specificity in the latter location [1,3]. The U-fibers may be involved in the anterior temporal and superior frontal regions [5]. Lacunar infarcts occur mostly in the semioval centre, thalami, basal ganglia and pons [3]. The number of lacunar infarctions is an important predictor of disability and cognitive decline [1,3]. Microbleeds appear as hypointense foci on T2-WI and even more pronounced on susceptibility weighted images (SWI) [3]. The diagnosis is made based on clinical and radiological features and genetic testing [1-3]. CADASIL may mimic multiple sclerosis (MS) both clinically and radiologically [4]. Radiological features which may help differentiation are contrast enhancement of active lesions in MS, absence of optic neuritis and of spinal cord involvement in CADASIL, sparing of the infratentorial brain, presence of microbleeds and typical bilateral anterior temporal lobe involvement in CADASIL [4]. Treatment consists of management of acute complications and strict blood pressure control [1,2]. In conclusion, the diagnosis of CADASIL should be suspected in recurrent ischaemic strokes in middle age adults without vascular risk factors, particularly if bilateral involvement of the anterior temporal lobes and insular regions is present. Written informed patient consent for publication has been obtained.

Differential Diagnosis List: CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy), Multiple sclerosis, Age-related and vascular risk-factor-related small vessel disease, MELAS, Fabry disease

Final Diagnosis: CADASIL (cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy)

References:

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Stojanov D, Aracki-Trenkic A, Vojinovic S, Ljubisavljevic S, Benedeto-Stojanov D, Tasic A, Vujnovic S (2015) Imaging characteristics of cerebral autosomal dominant arteriopathy with subcortical infarcts and leucoencephalopathy (CADASIL). Bosn J Basic Med Sci 15:1 (PMID: 25725137)
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Figure 1

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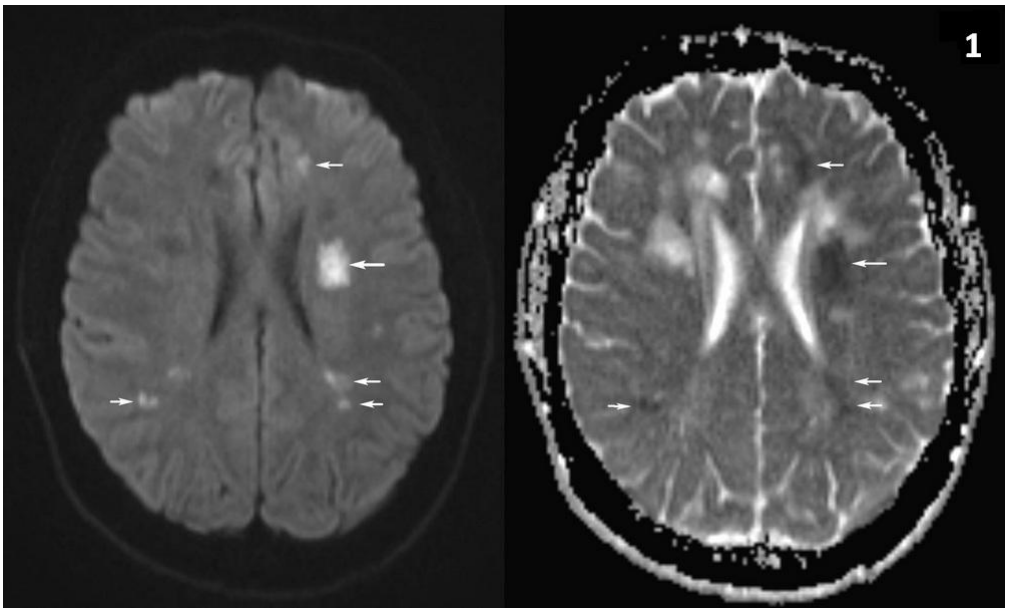

Description: MRI, axial diffusion weighted imaging (DWI) and apparent diffusion coefficient (ADC) map

Origin: Department of Radiology, AZ Sint-Maarten Mechelen 2800 Belgium, February 2019

Figure 2

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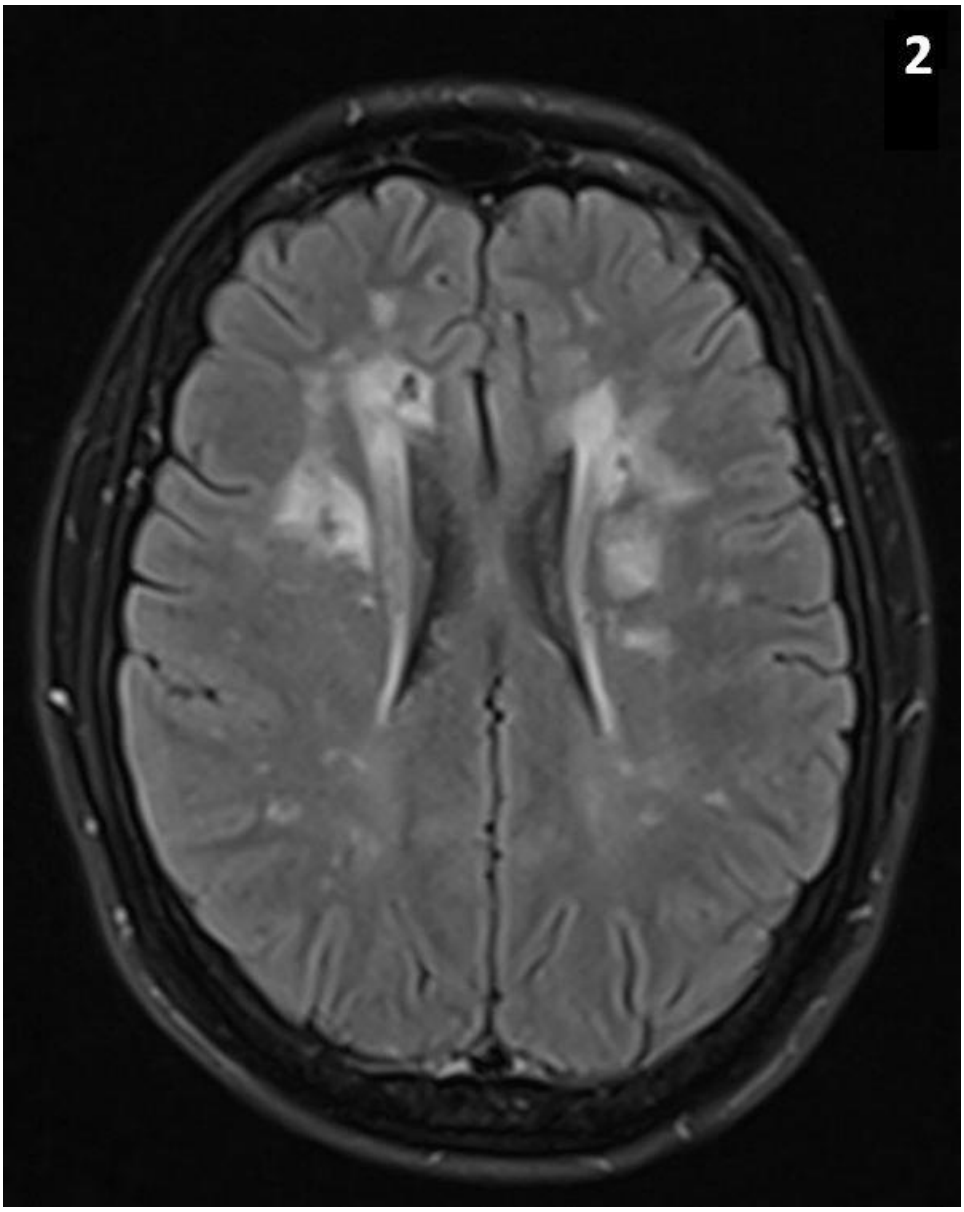

Description: MRI, axial FLAIR image **Origin:** Department of Radiology, AZ Sint-Maarten Mechelen 2800 Belgium, February 2019

Figure 3

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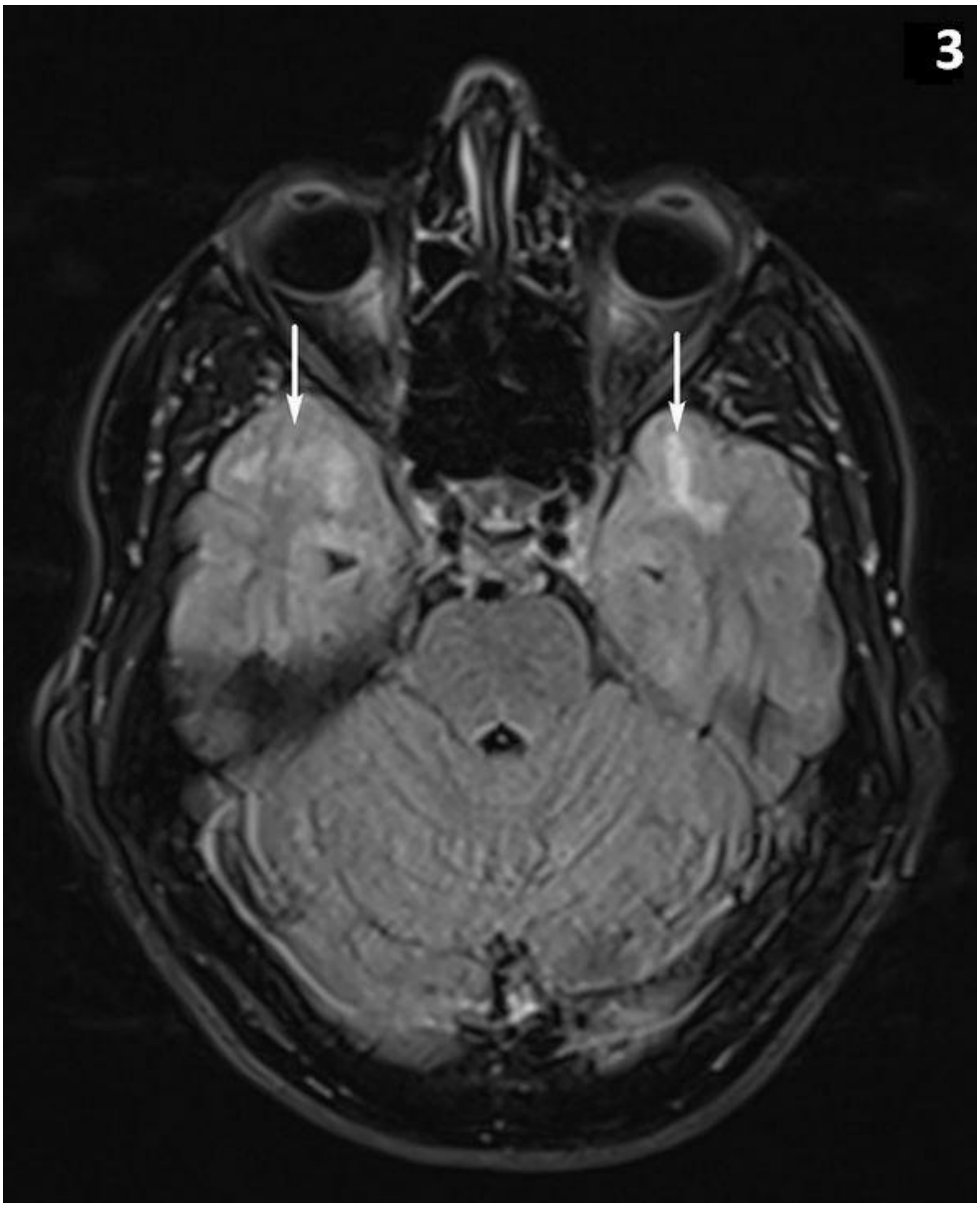

Description: MRI, axial FLAIR image **Origin:** Department of Radiology, AZ Sint-Maarten Mechelen 2800 Belgium, February 2019

Figure 4

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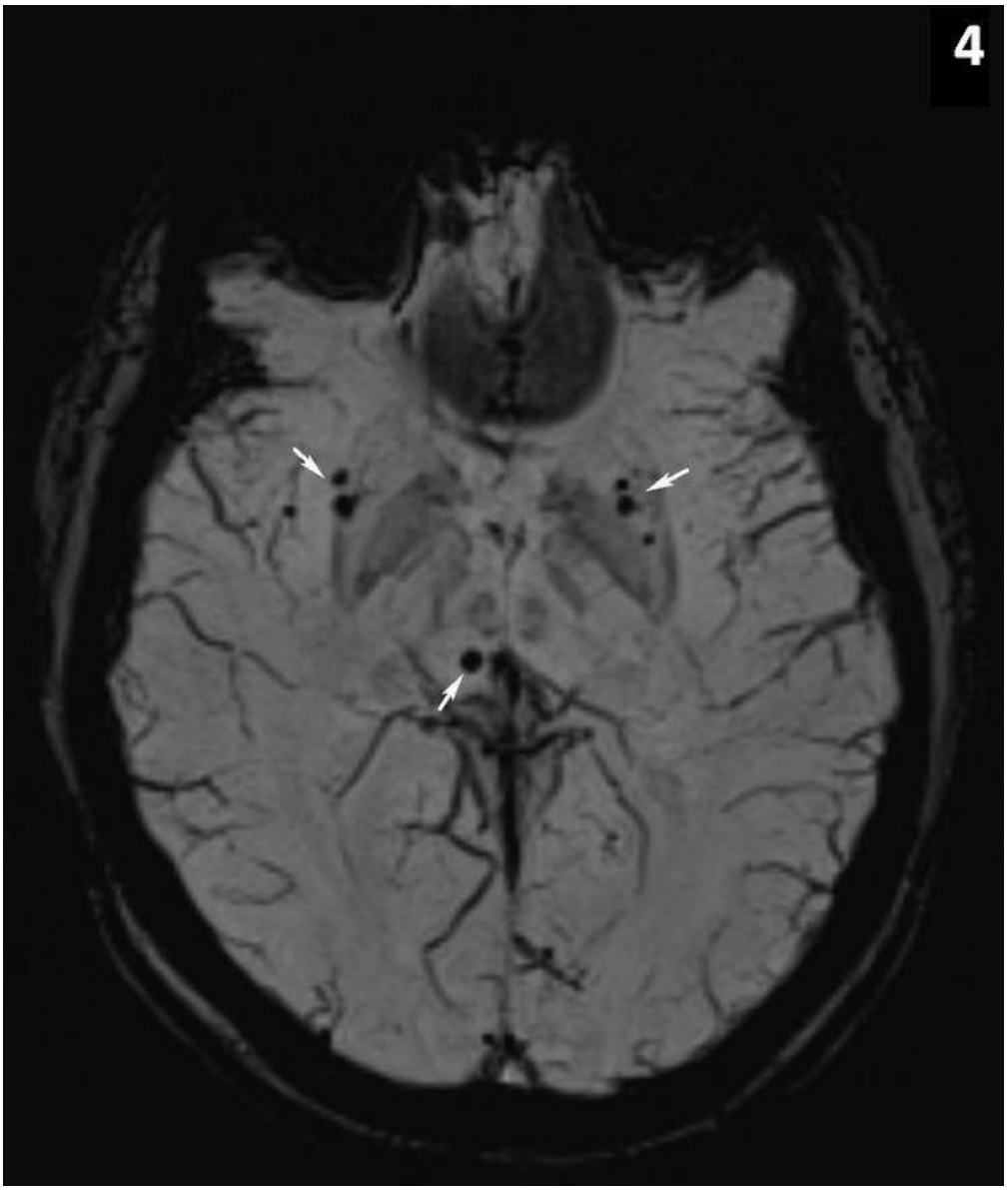

Description: MRI, axial SWI **Origin:** Department of Radiology, AZ Sint-Maarten Mechelen 2800 Belgium, February 2019