

# Bilateral internal carotid artery hypotrophy in malignant osteopetrosis

Ramli N, Rahmat K, Tan G P

## ABSTRACT

**Malignant osteopetrosis is associated with petrous carotid canal and internal carotid artery stenosis in the skull base. We present a four-year-old boy with malignant osteopetrosis who developed right frontal lobe infarction as a result of bilateral internal carotid artery hypotrophy.**

**Keywords: cerebral infarction, internal carotid artery hypotrophy, malignant osteopetrosis, osteopetrosis**

*Singapore Med J 2008;49(7):e175-e177*

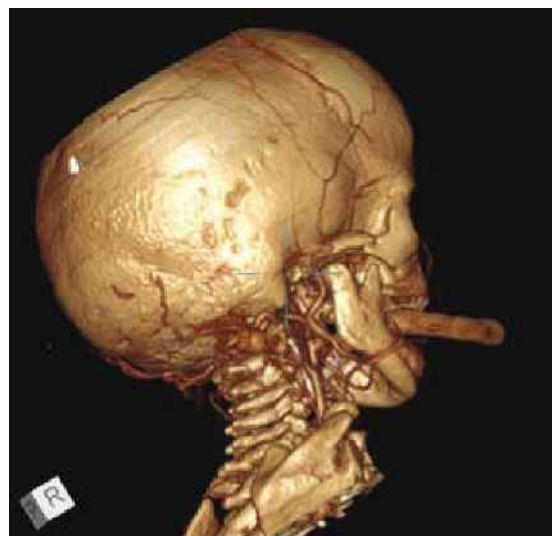
## INTRODUCTION

Petrous carotid canal (PCC) and internal carotid artery (ICA) stenosis within the skull base are common among patients with malignant osteopetrosis.<sup>(1)</sup> This cranial manifestation is due to the predominant calvarial bone thickening and poor osseous growth and remodelling. Cerebral ischaemia or infarction with osteopetrosis has rarely been reported.<sup>(2)</sup> We present a child with malignant osteopetrosis who developed right frontal lobe infarction as a result of bilateral internal carotid artery hypotrophy. Computed tomography (CT) angiogram and CT findings are discussed.

## CASE REPORT

A four-year-old boy was clinically diagnosed to have hereditary osteopetrosis at one year ten months of age. He previously had multiple blood transfusions for anaemia and had partial splenectomy due to hypersplenism. He suffered from hearing impairment and right upper motor neuron facial nerve palsy. His visual acuity was grossly normal. He had initially presented to our institution following a fall. He sustained a skull haematoma over the occipital region associated with generalised tonic-clonic fits.

Blood investigations showed anaemia with a haemoglobin level of 4.0 g/dL and thrombocytopenia with a platelet count of  $16 \times 10^9/L$ . He received packed cells and platelet transfusion. Non-enhanced CT revealed a small right frontal intraparenchymal haemorrhage, subarachnoid haemorrhage and non-communicating hydrocephalus. There was generalised thickening of the skull vault with non-pneumatised



**Fig. 1** Reconstructed 3D CT angiogram shows hypotrophy of the right internal carotid artery.



**Fig. 2** CT angiogram shows hypotrophy of the left internal carotid artery (white arrows) from the bifurcation to the foramen lacerum.

petrous temporal bone, narrowed optic foramina and internal auditory meati in keeping with osteopetrosis.

He underwent CT angiogram, which revealed attenuation of both the cervical ICAs from the bifurcation to the foramen lacerum (Figs. 1 & 2). There were collateral vessels seen along the path of the (extracranial) ICA. The carotid canal within the petrous bone also appeared narrowed (Fig. 3). However, both vertebral and basilar arteries appeared normal. The circle of Willis and its main branches were also normal. During this time, there was development of a new hypodense area in the right frontal region in keeping

Department of  
Biomedical Imaging,  
University of Malaya  
Medical Centre,  
Kuala Lumpur  
50603,  
Malaysia

Ramli N, FRCR  
Consultant  
Radiologist and  
Lecturer

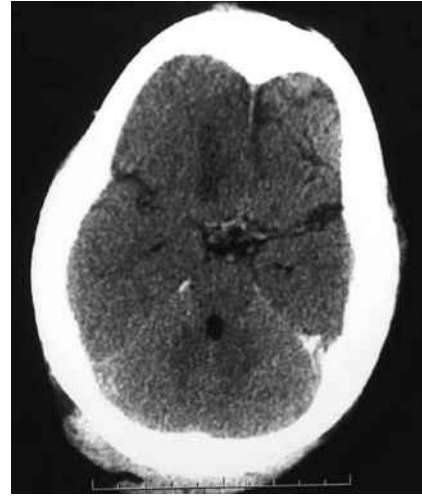
Rahmat K, FRCR  
Clinical Radiologist  
and Lecturer

Tan GP, MRad  
Clinical Radiologist

**Correspondence to:**  
Dr Kartini Rahmat  
Tel: (60) 3 7949 2069  
Fax: (60) 3 7958 1973  
Email: katt\_xr2000@  
yahoo.com



**Fig. 3** Axial CT image of the cranium shows a narrowed petrous carotid canal. Note the generalised thickening of the skull vault.



**Fig. 4** Axial CT image of the brain shows a hypodense area in the right frontal region, in keeping with cerebral infarction.

with cerebral infarction (Fig. 4). He was managed conservatively with blood product transfusion.

## DISCUSSION

Osteopetrosis is a rare metabolic bone disease caused by reduced osteoclastic activity, leading to deficiency in resorption of spongiosa and subsequent obliteration of medullary spaces associated with increased density of bone. Generally, there are two forms of the disease; the more malignant and often fatal autosomal recessive form (AROP), and the more benign autosomal dominant form (ADOP), which is often asymptomatic, commonly being discovered incidentally on plain radiograph. Clinically, osteopetrosis may present with anaemia, pathological fractures, neurological deficits and osteomyelitis. Radiographically, osteopetrosis are characterised by diffuse osteosclerosis. The greatest number of abnormalities occurs in patients with AROP, reflecting the greater severity of osseous abnormalities of this form of osteopetrosis. Cranial manifestations included thickening and sclerosis of the calvarial, ventriculomegaly, tonsillar herniation, proptosis and dural venous sinus stenosis in the majority of patients with AROP. Despite the frequency of venous stenosis in patients with AROP, venous infarcts were not observed.<sup>(3)</sup>

This patient demonstrated typical cranial CT findings in malignant osteopetrosis. His skull vault and base of skull were markedly thickened. There was narrowing of the optic foramina and the internal auditory meati. The petrous temporal bone was not aerated. Features seen most frequently, or exclusively, in AROP include stenosis of the ICA and vertebral artery, expansion of the subarachnoid spaces, brain atrophy, and extramedullary haematopoiesis.<sup>(3)</sup> Curé

et al showed that the funnel shape of most of the PCCs closely resembled the magnetic resonance (MR) angiographical shape of the petrous ICA in most of the patients, and hence derived that the petrous ICA stenosis identified on both conventional and MR angiograms in these patients are at least partially due to their small PCCs.<sup>(1,3)</sup> His study, which observed a positive correlation between PCC diameter and age, argues against progressive narrowing of the PCC.

Cerebrovascular complication is rarely reported. It has been postulated that physical inactivity of these children (a result of their anaemia, large, heavy head and propensity for fracture) may prevent a drastic challenge to their cardiac output and brain perfusion. Cerebrovascular complications can be either haemorrhagic or occlusive. Stenosis of ICA and vertebral artery are seen most prevalently, or exclusively in AROP.<sup>(4)</sup> This stenosis is postulated to be caused by narrowing of the PCC and transverse foramina due to osteopetrotic bone. Narrowing of the PCC and accompanied ICA stenosis are very common in AROP, but there is no correlation between the severity of carotid canal stenosis and age.<sup>(1)</sup> Very rarely, patients will suffer from ischaemic neurological complication. In two separate series reported by Curé et al with 34 and 20 patients, respectively, he observed that despite the high prevalence of venous and arterial stenosis, none of the patient suffered from cortical or venous infarct.<sup>(1,3)</sup> Wilm et al reported a 16-year-old boy who presented with transient ischaemic attack in the cortical middle cerebral artery territory on the right and stenosis of the vertebral artery, probably causing posterior fossa ischaemia with severe dizziness.<sup>(2)</sup> Other cerebrovascular manifestations include stenosis of jugular veins with narrowed jugular foramina.

This patient represents a more severe form of AROP affecting the vascular and skeletal system. Involvement of the proximal ICA vessels (as in this case), and the dural venous sinus in other reported cases, are likely due to retrograde hypotrophy. Lasjaunias et al mentioned that when narrowing is segmental (in this case, the entire ICA), then congenital hypoplasia is unlikely.<sup>(5)</sup> The presence of the carotid canal, although narrowed, also excludes a congenital origin. This patient demonstrated a never-before reported case of AROP with bilateral hypotrophy cervical ICAs, and as a result, developed the rare and unfortunate complication of cerebral infarction.

## REFERENCES

1. Curé JK, Key LL, Shankar L, Gross AJ. Petrous carotid canal stenosis in malignant osteopetrosis: CT documentation with MR angiographic correlation. *Radiology* 1996; 199:415-21.
2. Wilms G, Casaer P, Alliet P, et al. Cerebrovascular occlusive complications in osteopetrosis major. *Neuroradiology* 1990; 32:511-3.
3. Curé JK, Key LL, Goltra DD, VanTassel P. Cranial MR imaging of osteopetrosis. *AJNR* 2000; 21:1110-5.
4. Makin GJ, Coates RK, Pelz D, Drake CG, Barnett HJ. Major cerebral arterial and venous disease in osteopetrosis. *Stoke* 1986; 17:106-10.
5. Lasjaunias P, Berenstein A, Ter Brugge KG. *Surgical Neuroangiography*. 2nd ed. New York: Springer-Verlag, 2001: XXII.