

Commentary on “Neurological Manifestations as Presenting Feature of Osteopetrosis in Children: A Review of Eleven Cases”

Abbas Amirjamshidi^{1,*}

¹Sina Hospital, Tehran, IR Iran

*Corresponding author: Abbas Amirjamshidi, Sina Hospital, Emam Ave., Tehran, IR Iran, Tel.: +98-9121329864, Fax: +98-2177500958, E-mail: abamirjamshidi@yahoo.com.

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I read with interest the manuscript by Saidinia S et al. (1) about the neuro-manifestations of osteopetrosis. In these communications, the authors have succeeded in forwarding a well-developed discussion of different aspects of the illness and presented their experience with cases, referred to their Neurosurgical department and treated surgically by their group. Even though the information transferred by the authors is classically acceptable but there are delicate points to be reminded so that the readers might not be misled;

A) Although generally thought of as bone diseases, the most serious consequences of the Osteopetroses (OP) are seen in the nervous system in which the nerves, blood vessels and the spinal cord are compressed by either gradual occlusion or lack of growth of skull foramina. Most patients with OP have some degree of optic atrophy and many children with severe forms of autosomal recessive OP are rendered blind in whom; optic decompression is frequently attempted to improve their situation. Auditory, facial and trigeminal nerves may also be affected, and hydrocephalus can develop. Stenosis of both arterial supply (internal carotid and vertebral arteries) and venous drainage may occur but the least understood pattern for the disease is the neuronopathic OP. Approximately, 10% of infants have OP as part of a severe generalized neurometabolic disease. These occur to varying degrees depending upon the age of onset and severity of their disease. Accordingly, the exact mechanism of optic nerve damage is not entirely clear in all the patients. Although it is presumed that direct bony compression on the nerve is responsible, the nerve is not in direct contact with bone when assessed by CT scanning. This may be because the nerve has become atrophic and shrunk away from the bone. Regarding this, it is suggested that the attending

physician must consider very especial issues and variants before deciding for decompression of any cranial nerve in cases of OP (2-4).

B) The other potential mechanism leading in patients' complaints (not the shape to the skull) is the syndrome of raised pressure. In these patients, the stagnation of circulation occurs in the venules running over the surface of the optic nerve either by direct bony compression or by jugular venous outflow obstruction secondary to constriction of the jugular foramen. Either of these would produce back pressure, resulting in papilloedema and eventual optic atrophy.

Angiography may show severe narrowing of the internal carotid artery within the petrous carotid canal and stenosis of cervical vertebral arteries within the vertebral canal. More perplexingly, arteries can also be stenosed beyond these bony channels and stenoses of the supraclinoid portion of the carotid artery, and middle cerebral arterial occlusion have both been reported. Ventriculomegaly, tonsillar herniation, proptosis and dural venous sinus stenosis have been observed in magnetic resonance imaging (MRI) scans of a majority of OP patients. In severe cases of jugular foramen stenosis, the internal jugular vein can be occluded, leading to thrombosis of the transverse and sagittal sinuses. Shunting of blood from the superior sagittal sinus to the external jugular vein may explain the dilated scalp veins seen in some children. Acquired cephaloceles may also occur in OP, and Syringohydromyelia is also described. Enlargement of the optic canals has been demonstrated on computed tomography (CT) in some cases. Visual deterioration is usually stabilized but rarely improves, as many children have atrophic optic nerves by the time of treatment. Alternatively, increased intracranial pressure caused by foramen

Implication for health policy/practice/research/medical education:

Practice and research and education.

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magnum restriction could have the same effect. Optic nerve decompression by un-roofing of the orbit has been reported by a number of researchers. Visual improvement was reported in up to four of six operated nerves in some studies but other studies have reported variable results, including continued deterioration. It may be that the better results are obtained in those with the more benign forms of the disease versus the more malignant form (2, 3, 5, 6).

C) A number of researchers have reported primary retinal disease or central macular pigmentary changes with zones of geographical atrophy and mild optic disc pallor. It remains unclear whether all these have a variant of neuronopathic OP or whether several different diseases are involved (2, 4, 6).

D) Headache mostly appears later in life, and the mechanism is not clear. Raised intracranial pressure and hydrocephalus caused by impairment of the venous drainage of blood due to skull foramina could be a possible cause of it. Although successful un-roofing procedure of the orbit and other canals in the region of skull base has been reported by several authors, but the value and outcome of this procedure are of doubtful value. The ventricular and brain sizes are normal in most of the cases and the megalencephaly is either due to increasing skull thickness or retention of CSF after impaired venous drainage in any of the sinuses, jugular or the sagittal sinus. Lumbar puncture may reveal increased CSF pressure in some cases but the role and strict indications for inserting diversional systems for CSF or decompression of the foramen magnum are yet unclear (3-6).

We may conclude that it is not so easy to decide for any kind of neurosurgical intervention in cases affected by

OP, and the relevant literature lacks a well evidenced algorithm in this regard.

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Authors' Contribution

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