Rare case of dystrophia myotonica with mega cisterna magna

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CASE REPORT

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Abstract

Myotonic dystrophy is also known as dystrophia myotonica (DM). The condition is composed of at least two clinical disorders with overlapping phenotypes and distinct molecular genetic defects: myotonic dystrophy type 1, the classic disease originally described by Steinert, and myotonic dystrophy type 2, also called proximal myotonic myopathy (PROMM). Mega cisterna magna is thought to be an anatomic variant with no clinical significance. We report a rare case of type 1 dystrophia myotonica in combination with mega cisterna magna.

Key Words:
Dystrophia myotonica, mega cisterna magna, congenital myotonic dystrophy

Background

Myotonic disorders are those inherited muscle conditions characterised by slow relaxation due to spontaneous discharges. There are currently two clinical and molecular defined forms of myotonic dystrophy: 1) myotonic dystrophy type 1 (DM1), also known as 'Steinert's disease'; and 2) myotonic dystrophy type 2 (DM2), also known as proximal myotonic myopathy.[1] DM1 and DM2 are progressive multisystem genetic disorders with several clinical and genetic features in common. DM1 is the most common form of adult onset myotonic dystrophy whereas DM2 tends to have a milder phenotype with a later onset of symptoms and is rarer than DM1.[1]

The term mega cisterna magna has been loosely applied to an enlarged cisterna magna, also known as the cerebellomedullary cistern. Located between the posterior cerebellum and dorsal medullar oblongata, it has a greater than 10mm width with normal vermis and cerebellar hemispheres.[2]

We are reporting this rare case of dystrophia myotonica found in association with mega cisterna magna.

Case details

A 20-year-old patient presented to our outpatient medicine department with a history of vocal nasal twang. His speech was quite incomprehensible and the nasal twang increased on speaking loudly or on speaking continuously. In addition, once closed tightly, he had difficulty in opening his fists. To our knowledge, there was no family history of these symptoms.

On clinical examination, it was found that he had bradycardia (pulse: 58). The typical facial features of myopathy were present: elongated face, wasting of temporalis and bilateral sternocleidomastoids and frontal alopecia. (Figure 1) Bilateral hypothenar muscle wasting was also observed. Myotonia of distal muscles of both hands and percussion myotonia of tongue was present. Higher functions including mood, memory, and orientation were normal, although he had poor arithmetic calculating power. He had 9th and 10th cranial nerve palsy, characterised in this case by an absent gag reflex and vocal cord palsy.

Normal results were reported from baseline investigations, which included a complete blood count, random blood sugar, serum creatinine, serum electrolytes, serum thyroid stimulating hormone and chest x-ray. Muscle conduction studies indicated myotonia of left abductor pollicis brevis elicited spontaneously and percussively. MRI of the cranium revealed a mega cisterna magna. The patient was diagnosed with dystrophia myotonica type 1 in association with mega cistern magna. Phenytoin 300mg/day was given to the patient. With treatment, the patient showed partial improvement in myotonia.

Figure 1: A characteristic pattern of involvement of the facies in myotonia dystrophia.

Figure 2: Myotonic discharges on muscle conduction study

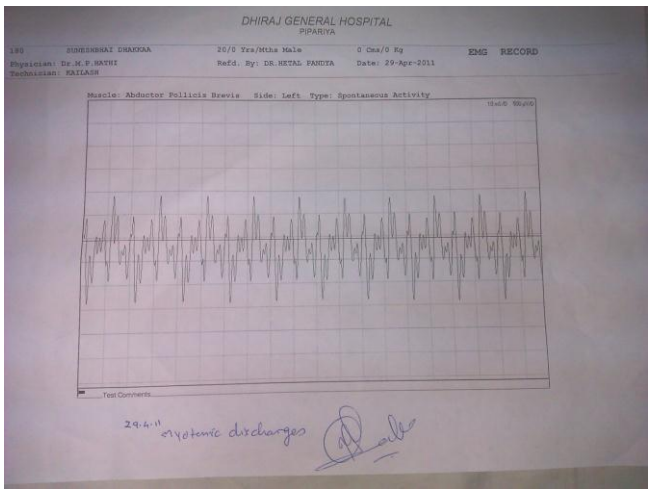
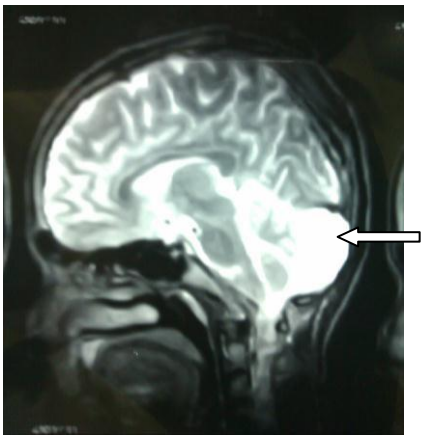


Figure 3: MRI cranium showing mega cisterna magna



Discussion

DM1 is the commonest cause of adult onset myotonic dystrophy. There is currently no cure but effective management is likely to significantly reduce the morbidity and mortality of patients. The enormous advances in the understanding of the molecular pathogenesis of DM1 and DM2 have resulted in the description of a novel disease mechanism involved in some myodegenerative and possibly neurodegenerative disorders.[1]

The onset of DM-1 occurs commonly in the second or third decade. However, the age of onset varies markedly with the generation affected. For example, in our patient, it occurred in the second decade, which may suggest the possibility that it is more closely related to a congenital variant and more severe form of DM1. This variant occurs in approximately 25% of infants of affected mothers and is characterised by severe facial and bulbar weakness, transient neonatal respiratory insufficiency, and mental retardation.[3, 4]

Various other congenital anomalies like hereditary motor and sensory neuropathy,[5] cerebral anomalies – ventricular dilatation, macrocephaly,[3] mitral valve prolapse[6] are reported to be associated with various types of myotonic dystrophy. We are reporting this rare case of myotonic dystrophy associated with another rare cranial anomaly – mega cisterna magna.

To evaluate the patient for nasal twang and lower cranial nerve palsy, the patient was investigated with an MRI brain study that also identified the presence of mega cisterna magna. At certain parts of the base of the brain, the arachnoid is separated from the pia mater by wide intervals, which communicate freely with each other and are named subarachnoid cisternæ; in these the subarachnoid tissue is

less abundant. The cisterna cerebellomedullaris (cisterna magna) is triangular on sagittal section, and results from the arachnoid bridging over the interval between the medulla oblongata and the under surfaces of the hemispheres of the cerebellum; it is continuous with the subarachnoid cavity of the medulla spinalis at the level of the foramen magnum.[7] Mega cisterna magna occurs in approximately 1% of all brains imaged postnatally and has been associated with cerebrovascular infarction, inflammation, and infection, particularly cytomegalovirus, as well as with chromosomal abnormalities, especially trisomy 18.[8] In the absence of other findings suggestive of a posterior fossa lesion, mega cisterna magna is unlikely to be clinically significant.[8] However, in this patient we found it in association with the adult variety of DM1.

As mega cisterna magna is reported in 1% of normal population postnatally[8] and 0.25%–0.33% of normal adult population[9,10] its association with myotonic dystrophy may be by chance. There are few studies postulating a relationship between CSF space abnormalities such as ventricular dilatation leading to hydrocephalus and congenital myotonic dystrophy.[3,11] Garcia-Alix and colleagues in 1991 reported the presence of ventricular dilatation evaluated by cranial ultrasonography in 11 out of 14 infants born with congenital myotonic dystrophy (78%).[3] They examined the brains of four of these cases who died and concluded that there was no ventricular obstruction although they observed a minor expression of neuronal migrational disturbance. They reported macrocephaly in 10 infants out of 17. In their study, they report a CSF space disturbance in a case of congenital myotonic dystrophy.[3] It is conceivable that – a variant of such pathology might have possible correlation with adult variety of DM1 in our patient. Although, to the best of our knowledge, this association is not yet described in medical literature and a large population-based study is a requirement for proving such association. A postulated reason might be that patients having congenital abnormality of CSF spaces may not be reaching adulthood, because of the possible development of serious complications like hydrocephalus. Minor CSF space abnormalities like dilatation of the cisterna, as observed in our patient, may not produce serious complications incompatible with reaching adulthood, and the patients having such abnormalities may remain asymptomatic and not investigated. As our patient had lower cranial nerve palsies, which was not possible to explain by DM1 only, the patient was further investigated with MRI of cranium and he was found to have mega cisterna magna.

The medical literature is not clear about the cause of ventricular dilatation in patients of DM1. Cerebral atrophy is a common accompaniment and ventricular dilatation is believed to be due to cerebral atrophy.[12] However, another hypothesis suggests that mega cisterna magna may originate during the foetal development process.[3]

Conclusion

This rare case of DM1 is reported in conjunction with the finding of mega cisterna magna. Whilst it remains unclear whether there is a relationship between these two findings in one patient, our coincidental observation, nevertheless, appears very rare.

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PEER REVIEW

CONFLICTS OF INTEREST

The authors declare that they have no competing interests

PATIENT CONSENT

Taken

The authors, Pandya H, Lakhani J, Mehta J, and Dodhania J, declare that:

1. They have obtained written, informed consent for the publication of the details relating to the patient(s) in this report.
2. All possible steps have been taken to safeguard the identity of the patient(s).
3. This submission is compliant with the requirements of local research ethics committees.