A case of Lhermitte–Duclos disease (LDD) (dysplastic gangliocytoma of cerebellum) associated with syringomyelia

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Background: Lhermitte–Duclos disease (LDD) (dysplastic gangliocytoma of cerebellum) is a rare hamartomatous lesion of cerebellar cortex. LDD may occur as a sporadic isolated disease or in association with a multiple hamartoma syndrome named Cowden Disease, that can be associated with a mutation in the PTEN gene on arm 10q (1). There are few cases of LDD associated with syringomyelia described in literature (2).

Case report: in April 2017, a 52-year-old man was referred to our institute with a 1-month history of dizziness and gait ataxia. He had been successfully treated with steroids for a right facial peripheral nerve palsy approximately four months earlier. His medical anamnesis revealed multinodular goiter, previous embolization of congenital arteriovenous fistula of the hand and removal of renal cell carcinoma and cutaneous fibropapillomas.
Brain MRI showed a partial calcified lesion involving the right cerebellar hemisphere and the vermis.
The lesion had a characteristic gyriform pattern, which was hypointense on T1W and hyperintense on T2W sequences, giving it a classical tigroid appearance.
On DWI a bright signal was noted, whereas there was no disturbance on ADC maps. MR Spectroscopy revealed elevated lactate, mildly decreased Cho/Cr and normal NAA/Cr ratio. There was tonsillar herniation with obstructive hydrocephalus and syringomyelia of the cervical cord from C2 to C5 level. The patient could not receive gadolinium due to a previous severe adverse reaction.

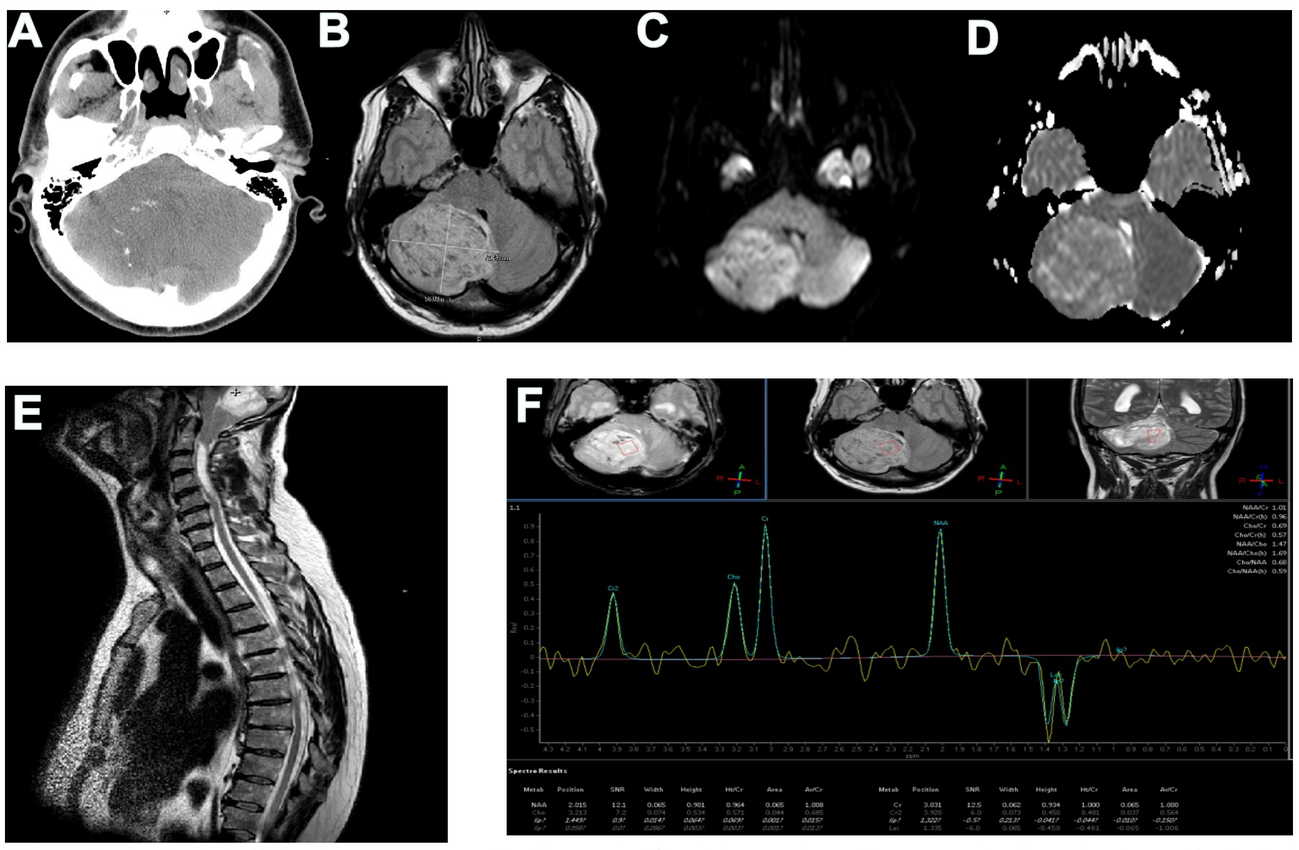

Fig. 1 A *Axial CT shows a partially calcified low density cerebellar lesion* ***B*** *Axial T2-weighted MRI shows the thickened high signal folia, characteristic of LDD* ***C,D*** *On DWI a bright signal was noted, whereas there was no disturbance on ADC maps* ***E*** *T2-weighted midsagittal image of the cervicodorsal spine shows the syrinx* ***F*** *MRS (TE 135 ms) shows the presence of lactate (arrow).*

He underwent EVD placement followed by right suboccipital craniectomy, C1 arch excision and decompression of the tumor.

At surgery the right cerebellar hemisphere appeared markedly thickened and enlarged and the tumour showed high vascularization. A macroscopically complete resection of the lesion was performed. Histopathological examination revealed replacement and expansion of the internal granular cell layer by large neurones with vesicular nuclei and prominent nucleoli. The features were consistent with LDD.

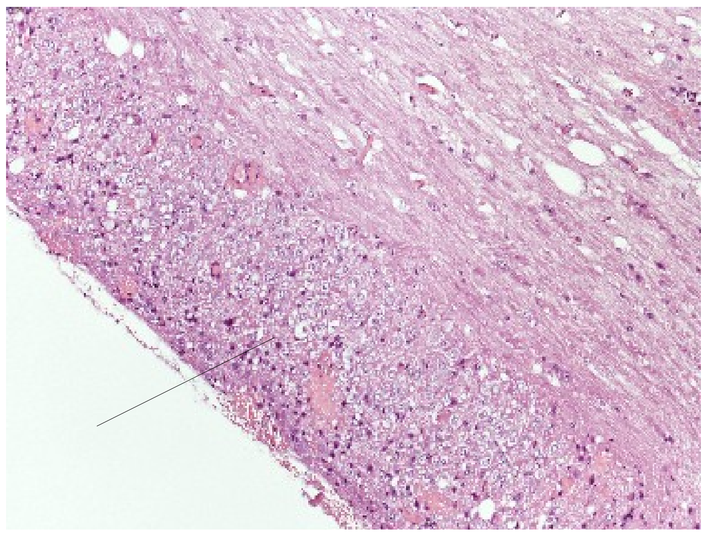

Fig. 2 Histology*: the granule cells are totally replaced with abnormal large ganglion cells (arrow). Haematoxylin and eosin (x10).*

The postoperative course was complicated by decreased vigilance and acute respiratory failure. Brain CT scan showed bleeding within the surgical cavity and fibrobronchoscopy revealed dense tracheobronchial secretions and fibroepithelial polyps which were removed. Tracheostomy was placed and the patient's condition gradually improved. He has been recently transferred to rehabilitation.

Conclusion and discussion: we report a rare case of LDD associated with syringomyelia. Cerebellar tonsillar herniation probably acts as precursor of formation of syrinx in this condition (2). A Cowden syndrome may also be suspected based on the medical history of this patient, but it will have to be confirmed by mutational analysis of the PTEN gene.

References

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