CASE REPORT

Lhermitte-Duclos disease associated to Cowden syndrome: de novo diagnosis and management of these extremely rare syndromes in a patient

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SUMMARY

A 36-year-old woman, with history of cutaneous papilomatosis and thyroid carcinoma presented with headache, transitory visual blurring and nausea. Funduscopy showed papilloedema. MRI showed a tumour of the right cerebellar hemisphere with a striated, tigroid pattern, typical of Lhermitte-Duclos disease (LDD). Significant clinical and perimetric improvements were noted after surgery and the follow-up did not reveal recurrences of the tumour. LDD is an extremely rare differential diagnosis of posterior fossa tumours. LDD and the history of thyroid carcinoma permitted us to diagnose Cowden syndrome (CS). We present a clinical case that supports the possibility of performing a preoperative diagnosis of LDD based on MRI features. We review the diagnosis and management of LDD and CS. This report highlights the importance of excluding CS after LDD diagnosis, of monitoring the optic nerve postoperatively using optical coherence tomography and of prompt treatment that can potentially prevent visual function loss.

BACKGROUND

Cowden syndrome (CS) is an extremely rare phacomatosis.[1–12] CS was first documented in 1963 by Lloyd and Dennis and it is a rare autosomal dominant multisystem syndrome with incomplete penetrance and variable expressivity, involving hamartomatous overgrowth of tissues of all three embryonic origins and predisposing to multiples cancers.[2–9] Eighty per cent of CS cases are caused by a germline mutation in the *PTEN* gene (*phosphatase and tensin homologue*), which is a tumour suppressor gene located on chromosome 10q22–23.[10] CS is associated with an increased risk of breast, thyroid and endometrial cancer development.[5–9] CS is characterised by pathognomonic mucocutaneous abnormalities, such as multiple papulous and papilomatous lesions of the skin or mucous membranes, facial trichilemmomas, acral keratosis and also by multiple hamartomatous neoplasms of the skin, oral mucosa, gastrointestinal tract, bones, breast, thyroid, eyes and genitourinary tract and central nervous system (CNS).[9 11 12] In the CNS, the cerebellum can also be affected by this hamartomatous growth. Lhermitte-Duclos disease (LDD) is an extremely rare clinical entity and it is characterised by a unilateral cerebellar hamartoma, a dysplasic gangliocytoma (DGC), with slow progression, which can cause an intracranial mass effect with occlusive hydrocephalus, needing urgent surgical intervention.[2 10 13–17] LDD is a major criterion for the diagnosis of CS.[12 18] From 1963 to 2006, only 220 cases of LDS were described worldwide.[17] LDD is an extremely rare differential diagnosis of posterior fossa space occupying lesions and it is a major manifestation of the rare CS.[1 2 11 12] Our case supports the possibility of performing a preoperative diagnosis of LDD based on pathognomonic MRI findings. The knowledge of the pathognomonic feature of LDD on MRI is fundamental for an early preoperative diagnosis. The purpose is to highlight the MRI features of LDD that permit a preoperative diagnosis, to review the diagnosis and management of LDD, with literature review and the report of an illustrative clinical case. This report also highlights the importance of a prompt treatment which can potentially prevent visual function loss, the role of optical coherence tomography (OCT) as a useful tool to monitor the oedematous status of the optic nerve after surgery and the need to exclude CS and associated malignancies after LDD diagnosis.

CASE PRESENTATION

A 36-year-old woman came to emergency room for headache with 2-week duration, associated to visual blurring bilaterally (OU). She also reported nausea, but denied loss of consciousness or vomiting. She had history of several papillomatous lesions of the skin that were surgically removed and history of total thyroidectomy for a thyroid carcinoma and of fibrocystic lesions in the breasts. Relatively to her family history, her mother and sister had breast carcinoma, treated by total mastectomy and chemotherapy and radiotherapy.

The best-visual acuity was 20/20 in right (OD) and left (OS) eyes, intraocular pressure was 15 mm Hg by Goldmann applanation tonometry OU. Both pupils were equal in size and the pupillary light reflexes were symmetrical. Ocular movements were normal. Pain and diplopia were absent. Visual field testing by confrontation and biomicroscopy were unremarkable OU. The funduscopy showed papilloedema (figure 1A). CT of the head showed a unilateral isoattenuated cerebellar mass in the right hemisphere (figure 1B) and obstructive hydrocephalus (figure 1C). Nuclear MRI showed a right cerebellar mass with secondary hypertrophy of the cerebellar folia and with a striated or tigroid pattern characteristic of LDD (figures 2A–D). This right-sided folial hypertrophy caused a marked asymmetry between cerebellar hemispheres.

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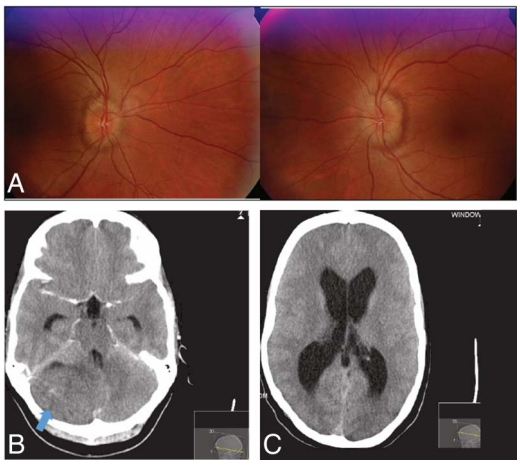

Figure 1 Retinography (A) of both eyes showing papiloedema (Modified Frisén Scale grade 2). Unenhanced CT (B) showing a posterior fossa tumour in the right cerebellar hemisphere (blue arrow) with isoattenuation, causing significant mass effect causing partial collapse of the fourth ventricle (arrow). Supratentorial ventriculomegaly was present (C).

These findings permitted a preoperative diagnosis of LDD. The neurological examination did not show any other abnormalities. Computerised static perimetry (CSP) with a commercial perimeter (Octopus 900, Haag-Streit Diagnostics, Switzerland), using tendency-oriented perimetry strategy (G2-program) showed inferior arcuate scotomas in OU and a central scotoma in OS (figure 3A). OCT (Spectralis, Heidelberg Engineering, Heidelberg, Germany) showed a generalised thickening of the peripapillary retinal nerve fibre layer (RNFL) OU (figure 3B). OCT in enhanced depth imaging (EDI) mode showed an elevation of the optic nerve head OU with a prominent subretinal hyporeflective space with a relatively regular internal lining, compatible with papilloedema (figure 3C). Fluorescein angiography (FA) showed late leakage from the optic discs and confirmed papilloedema (figure 3D).

TREATMENT

The patient was admitted to hospital for surgical treatment and inpatient treatment with intravenous mannitol (substituted to furosemide 60 mg/day after the first 12 hours), and dexamethasone (10 mg q8h) was initiated. A ventriculoperitoneal shunt was placed the following day and the cerebellar lesion was surgically removed 1 week later.

OUTCOME AND FOLLOW-UP

Significant clinical and perimetric improvements occurred 2 weeks after surgery. The genetic test revealed *PTEN* mutation c.493G>T. No recurrences of the papilloedema occurred neither any new intracranial mass was seen on neuroimaging during follow-up until 12 months after surgery. At this time, the RNFL thickness (RNFLT) was within reference values in all peripapillary locations (figure 4A) and CSP did not show significant abnormalities

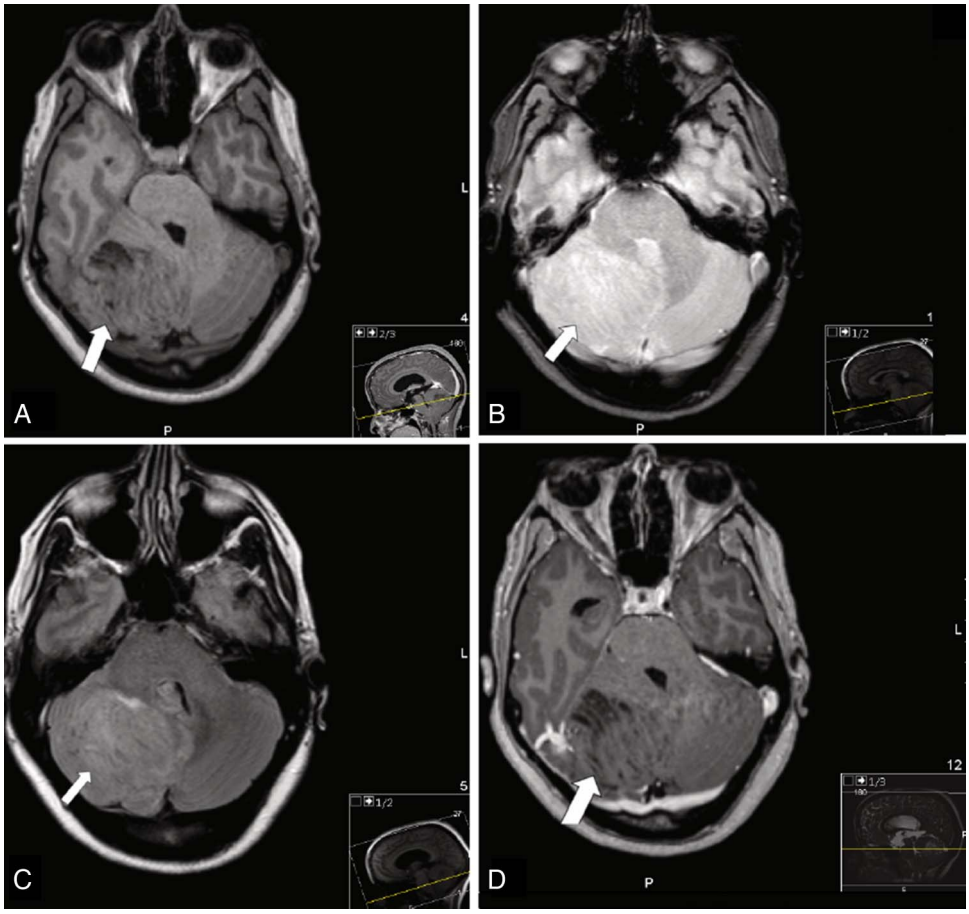

Figure 2 Axial MRI presented the typical striated appearance, with alternating hypointense and isointense bands (arrow) in T1-weightened (T1-W) images (A) and alternating high-intensity and normal-signal-intensity bands (arrow) in T2-weightened (T2-W) images (B). Axial fluid-attenuated inversion-recovery (FLAIR) MRI (C) showed global hyperintensity of the lesion (arrow). Axial T1-W-MRI with intravenous gadolinium (D) showing no enhancement of the cerebellar tumour (arrow). The typical striated pattern on T1-W images is preserved and well demonstrated here.

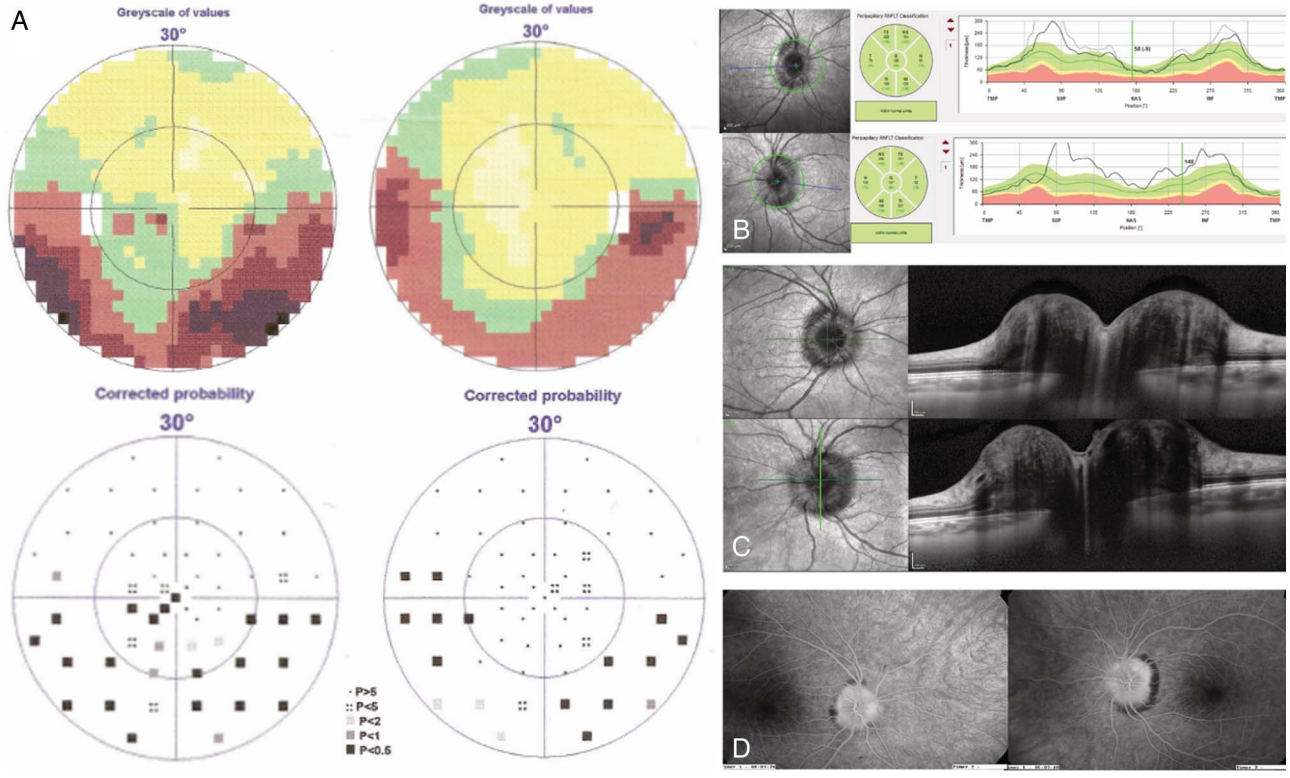

Figure 3 Computerised static perimetry (Octopus 900, Haag-Streit Diagnostics, Switzerland) of the right eye (OD, at the right) and left eye (OS, at the left) with tendency-oriented perimetry strategy (G2 program) showing dense inferior arcuate scotomas in OD and OS and a central scotoma in OS (A). The mean deviation (MD) was 4,7 dB in OD and 7,3 dB in OS. The loss variance (LV) was 27,0 dB in OD and 50,5 dB in OS. Spectral-domain optical coherence tomography (SD-OCT) of the right (top) and left (bottom) eyes showed increased retinal nerve fibre layer thickness in all peripapillary locations bilaterally (B). SD-OCT on Enhanced Depth Imaging mode of the right (top) and left (bottom) eyes showed characteristic features of papilledema (C), such as raised optic nerve head (ONH) surfaces with increased subretinal hyporeflective spaces (SHYPS), which had a smooth internal lining, as a recumbent 'lazy V' pattern of the SHYPS. Fluorescein angiography (D) demonstrated bilateral late leakage at the optic nerve in both eyes, which is a characteristic of papilloedema.

(figure 4B). The screening for breast, endometrial and renal malignancies were negative. The patient will be maintained in a regular follow-up programme, with evaluations every 6 months.

DISCUSSION

Patients with LDD should be evaluated carefully for signs of CS (box 1 and table 1). As CS is extremely rare, our knowledge of this syndrome is based mainly on compilations of case reports.[18] CS is frequently an easily missed diagnosis, because of the variability of the presentation and the rareness of the disease. Most cases are diagnosed in an advanced stage, when significant manifestations are present, as occurred in our case.[2] According to the *International Cowden Consortium* operational criteria for the diagnosis of CS (box 1), LDD is considered a major manifestation of CS and a major criterion for its diagnosis.[19] Other major criteria include breast or thyroid carcinomas and macrocephaly.[19] The presence of two major criteria in our patient, such as LDD and thyroid carcinoma made the diagnosis of CS, which was corroborated by the presence of pathognomonic skin lesions and a positive genetic test for a *PTEN* mutation.[19] The Cleveland Clinic (CC) Adult Score System (table 1) was created to identify candidates for *PTEN* testing, which is recommended for CC scores ≥10.[20] The diagnosis of LDD alone is classified with a score of 10. Our patient had a total CC score of 15, which justified the *PTEN* testing.

LDD is a focally indolent growth of the cerebellar cortex in which the folia enlarge due to a profusion of dysplastic cortical neurons and a thickening of the molecular layer.[21–23] There is loss of Purkinje cells and thinning of medullary white matter occur in this process.[21–23] The enlarged folia lose their secondary foldings and asymmetrically expand the cerebellar hemisphere.[21 23] These morphological features produce a characteristic pattern MRI, affording an opportunity for a preoperative diagnosis.[21 23] LDD is an imaging-specific diagnosis because of its typical striated folial pattern on MRI that consists of alternating bands on both T1-weighted and T2-weighted images.[21–23] The bands are hyperintense and isointense relative to grey matter on T2-weighted images and isointense and hypointense on T1-weighted images, which represents the abnormally thickened folia.[21–23] The tumour is characteristically not suppressed on axial fluid-attenuated inversion-recovery (FLAIR) MRI.[22] In our case, the mass was hyperintense on FLAIR-MRI. Calcification is an uncommon finding, but it was absent in our patient.[22 23] Most DGC do not enhance, as in our case; however, enhancement has been reported.[24] It has been suggested that MRI is sufficient for the diagnosis of this condition.[21 23 25] Typical MRI features of LDD are summarised on box 2. Most DGC are hypoattenuated on unenhanced CT; however, they can also be isoattenuated as in our case, with almost imperceptible borders.[22]

Mass effect is common and causes compression of the fourth ventricle and occlusive hydrocephalus, which was responsible for the symptoms and funduscopic findings in our patient.[2 22] Malignant transformation has not been reported.[2] LDD is seen most frequently in young adults (third and fourth decades) and there is no sex predilection.[22 26] Clinically, patients may be

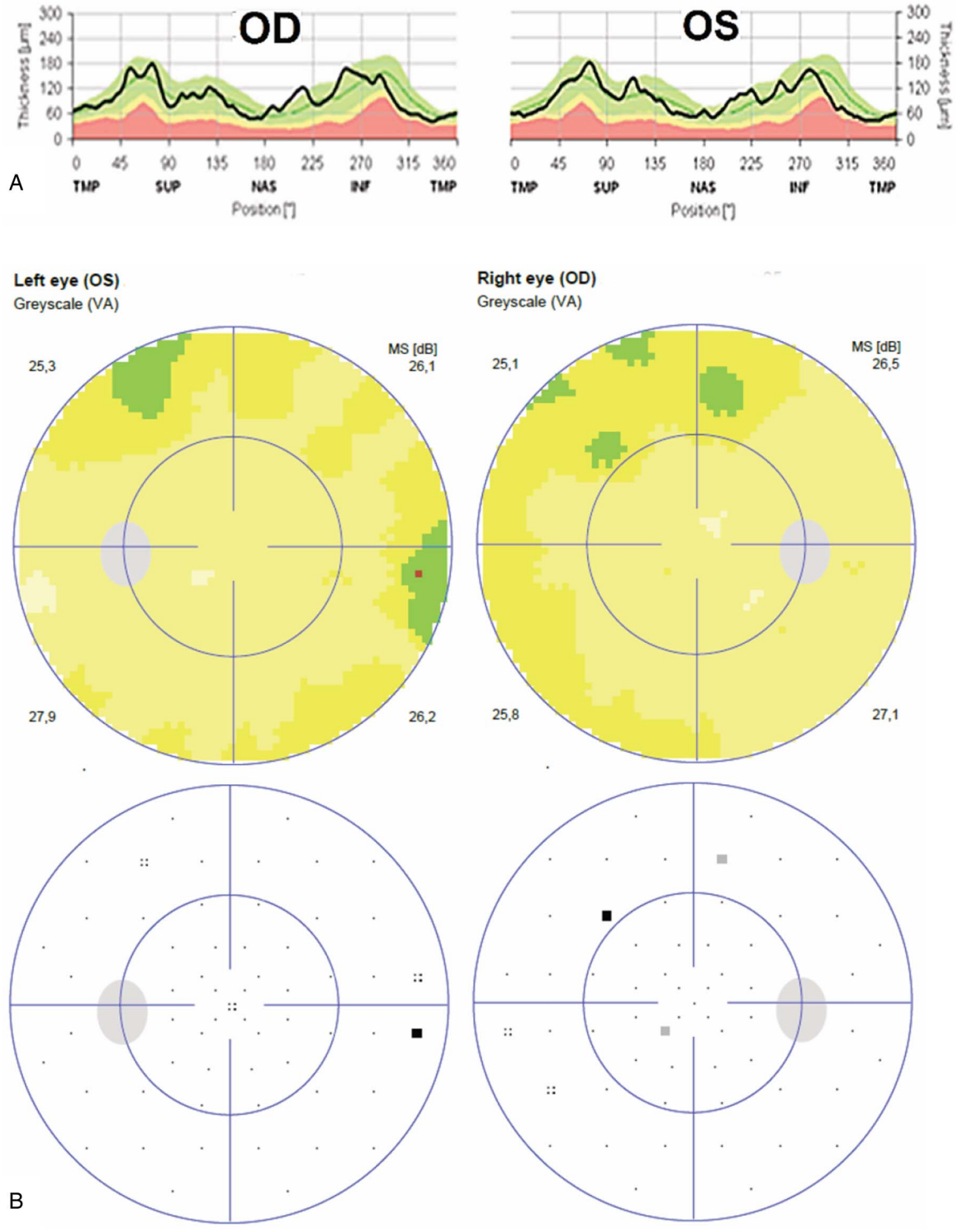

Figure 4 At 1 year after surgical removal of the cerebellar tumour, peripapillary retinal nerve fibre layer thickness of the right (OD) and left (OS) eyes was normal in all locations (A) and visual field testing (Tendency-oriented perimetry, G2 program, Octopus 900, Haag-Streit Diagnostics, Switzerland) did not reveal significant perimetric abnormalities (B). Mean deviation (MD) was 2,3 dB in OD and 2,5 dB in OS. Squared root of the loss variance (sLV) was 2,6 in OD and 2,4 in OS.

asymptomatic, or, when the lesion growths significantly, they may present with symptoms and signs of increased intracranial pressure, as in our patient.[16 22] Cranial nerve palsies, cerebellar symptoms, can occur (table 2), but were absent in the reported case.[16] This disease is commonly associated with other congenital malformations, such as macrocephaly, polydactyly, multiple haemangiomata and skull abnormalities, but these were also absent in our patient.[11] Possible clinical manifestations of sporadic and CS-related LDD are summarised on table 2. In our case, a preoperative diagnosis of LDD was made based on the presence of the pathognomonic features on MRI (box 2). This diagnosis was also supported by the presence of the *PTEN* mutation and later confirmed by pathological examination of the excised tumour. Our case supports the possibility of a

Table 1 Cleveland Clinic Adult Scoring System to identify candidates for PTEN testing

CC Adult Scoring System Manifestations	Score
Neurological	
Macrocephaly	6
Lhermitte-Duclos disease	*10**
Autism or developmental delay	1
Breast and gynaecological	
Invasive breast cancer at age <40 years	4
Invasive breast cancer at age between 40 and 49 years	2
Fibrocystic breast disease	1
Endometrial cancer at age <30 years	10
Endometrial cancer at age 30–49 years	6
Endometrial cancer at age ≥50 years	1
Fibroids	1
Gastrointestinal	
Polyposis syndrome (≥5, any type)	6
Intestinal hamartoma or ganglioneuroma, any number	10
Glycogenic acanthosis	10
Skin	
Trichilemmomas, biopsy proven	10
Oral papillomas	6
Penile freckling	6
Acral keratoses	1
Arteriovenous malformations	6
Skin lipomas	1
Endocrine	
Thyroid cancer at age <20 years	10
Thyroid cancer at age <50 years	4
Thyroid cancer at age ≥50 years	1
Thyroid goitre, nodules, adenomas or Hashimoto's thyroiditis (one or more features)	4
Genitourinary	
Renal cell carcinoma	1
An online CC score calculator is available at http://www.lerner.ccf.org/gmi/ccscore/	

**PTEN* mutation testing is recommended for a total CC score ≥10.
[20]Tan, *et al*. A Clinical Scoring System for Selection of Patients for PTEN Mutation Testing is Proposed on the Basis of a Prospective Study of 3042 Probands. Am J Hum Gen 2011;88(1):42–56.
CC, Cleveland Clinic.

Box 1 Diagnostic criteria of Cowden syndrome (CS)

International Cowden Consortium (ICC) operational criteria for the diagnosis of Cowden syndrome (CS)

Patognomonic criteria

Mucocutaneous lesions

- Trichilemmomas
- Acral keratosis
- **Papillomatous papules***
- Mucosal lesions

Major criteria

Lhermitte-Duclos Disease (LDD)*

- Breast carcinoma
- **Thyroid carcinoma*** (non-medullary), especially folicular thyroid carcinoma
- Macrocephaly (megalencephaly) (say, ≥95th centile)
- Endometrial carcinoma

Minor criteria

Other thyroid lesions (eg, adenoma or multinodular goitre)
Mental retardation (say, intelligence quotient (IQ) ≤75)
Gastrointestinal hamartomas
Fibrocystic disease of the breast*

- Lipomas
- Fibromas
- Genitourinary tumours (eg, renal cell carcinoma, uterine fibroids) or malformation.

Operational diagnosis

1. Mucocutaneous lesions alone if:
   A. There are 6 or more facial papules, of which 3 or more must be trichilemmoma, or
   B. Cutaneous facial papules and oral mucosal papillomatosis, or
   C. Oral mucosal papillomatosis and acral keratoses, or
   D. Palmoplantar keratoses, 6 or more
2. **2 major criteria but one must include LDD or macrocephaly**
3. 1 major and 3 minor criteria
4. 4 minor criteria

Operational diagnosis in a family where one person is diagnostic for Cowden syndrome (CS)

1. The pathognomonic criterion/ia
2. Any one major criterion with or without minor criteria
3. Two minor criteria

***Criteria present in our patient.**

Source: Pilarski and Eng[12]

preoperative diagnosis based on MRI, as indicated in the literature.[21–23]

Peripapillary RNFL imaging by OCT allows monitoring the effect of surgical removal of the tumour and lowering of intracranial pressure on the oedematous status of the optic nerve and peripapillary retina and for this reason baseline imaging is important. With the advent of EDI-OCT, new features of optic disc oedema were described, such as a raised optic nerve head (ONH) surface with an increased subretinal hyporeflective space (SHYPS), which had a smooth internal lining, as a recumbent 'lazy V' pattern of the SHYPS.[27] Our case presented with typical features of papilloedema on EDI-OCT. Short-term follow-up showed a favourable clinical course with normalisation of the RNFLT after surgery and long-term follow-up showed the absence of significant RNFLT defects. Findings on FA also supported the funduscopic diagnosis of papilloedema.

Obstructive hydrocephalus is associated with several visual field defects.[28] Even with surgical correction of the cause, significant visual sequela can persist.[28] Fortunately, this was not the case in our patient, probably due to the shorter duration of papilloedema and intracranial hypertension. The immediate goal of treatment of LDD is to decompress the ventricular system in symptomatic patients.[22 29] A ventricular shunt is placed initially, which is followed by tumour resection. According to the literature, the postoperative period is usually uneventful, as occurred in our case.[16] However, complete resection may be difficult because of poorly defined margins, leading to a high risk of recurrence. For this reason and given the slowly-growing nature of LDD, a long-term follow-up is required.[30 31] However, our case highlights that prompt and aggressive surgical treatment can prevent significant visual loss, normalise the peripapillary retinal nerve fibre layer and preserve the visual field.

Table 2 Clinical presentation of sporadic and Cowden syndrome-related Lhermitte-Duclos disease

Clinical presentation of Lhermitte-Duclos disease	
Sporadic LDD (Clinical findings of LDD in the absence of systemic manifestations of CS)	**CS-related LDD (clinical findings of LDD and systemic features of CS)**
Clinical presentation	Clinical presentation
Asymptomatic	The same of sporadic LDD
Symptoms and/or signs of intracranial hypertension	
Cranial nerve palsies (30%)	
Cerebellar dysfunction (40–50%): ataxia, dysdiadochokinesia, …	
Less common: loss of consciousness, seizures, neck stiffness, tinnitus.	
Ophthalmological features	Ophthalmological features
Decreased BCVA	The same of sporadic LDD
Visual field defects (central, centrocentral, blind spot enlargement, arcuate, …)	Ophthalmological features of CS can also be present:
Papilloedema (intracranial hypertension): ▶ Funduscopy—optic discs with blurred borders and non-visible cups. ▶ OCT shows increased RNFLT in all peripapillary quadrants. ▶ EDI-OCT—shows elevation of optic nerve heads, increased SHYPS with a regular internal lining and a recumbent 'lazy V' pattern of the SHYPS. ▶ FA—bilateral optic disc hyperfluorescence with late leakage.	▶ Optic nerve head drusen; ▶ Iris mamillations; ▶ Typical CS skin lesions occurring in periocular tissues/eyelids; ▶ Conjunctival papillomas; ▶ Proliferative retinopathy; ▶ Uveitis; ▶ Glaucoma; ▶ Combined hamartoma of retina and retinal pigment epithelium.
Other features:	Other features:
Megalencephaly, polydactyly, multiple haemangioma and skull abnormalities, mental retardation, other CNS malformations: hydromyelia, brain heterotopia and megalencephaly	The same of sporadic LDD plus systemic manifestations of CS (see box 1 and table 1)

BCVA, best-corrected visual acuity; CNS, Central Nervous System; CS, Cowden Syndrome; EDI, Enhanced Depth Imaging; FA, fluorescein angiography; LDD, Lhermitte-Duclos Disease; OCT, optical coherence tomography; RNFLT, peripapillary retinal nerve fibre layer thickness; SHYPS, subretinal hyporeflective space.

Box 2 Neuroimagiological findings of Lhermitte-Duclos disease on MRI in adults

Neuroimaging findings in adults

Magnetic resonance imaging (MRI) findings suggesting Lhermitte-Duclos disease (LDD)

- Cerebellar lesion in a patient with Cowden syndrome
- Slowly-growing cerebellar lesion on follow-up MRI scans
- Occlusive hydrocephalus
- Unilateral hemispheric expansion
- Preservation of the gyral pattern

MRI findings permitting a preoperative diagnosis of LDD

- **-Cerebellar tumour with a typical striated pattern that respects the cerebellar convolutions despite its enlargement**
- **-A 'tiger-striped' cerebellar lesion**
- **T1W:** predominant hypotensive lesion with a striated/tygroid pattern with alternated hypointense and iso-intense streaks
- **T2W:** —predominant hyperintense lesion with a striated/tygroid pattern with alternating hyperintense and iso-intense streaks.
- **FLAIR—**does not suppress the lesion
- Non-enhancement after gadolinium intravenous injection (most cases)

MRI findings atypical for LDD

- Enhancement after gadolinium intravenous injection (however it can occur in LDD)

Not suggestive of LDD

- Bilateral cerebellar lesions
- Multiple cerebellar lesions
- Homogenous pattern on T1 and T2
- Absence of striated pattern of the lesion on T1 and T2 in adults (but LDD without the typical striated pattern can occur in children)
- Predominantly hyperintense lesion on T1
- Predominantly hypointense lesion on T2

The diagnosis of LDD obligates to screen for systemic features of CS presented in box 1 and table 1, given the high association between both syndromes.

LDD, Lhermitte-Duclos disease; CS, Cowden syndrome; FLAIR, fluid-attenuated inversion-recovery MRI, T1W, T1-weighted MRI; T2W, T2-weighted MRI.

Table 3 Differential diagnosis based on clinical and neuroimagiological findings

Clinical and MRI findings	Comment
Papilloedema	Intracranial hypertension
Dilation of aqueduct of Sylvius, compression of fourth ventricle, triventricular hydrocephalus	Obstructive hydrocephalus secondary to mass effect produced by the cerebellar lesion.
Lack of enhancement	Helped to rule out leptomeningeal metastases and cerebellar encephalitis
Typical striated appearance of LDD	Permits the preoperative diagnosis of LDD in an adult patient. In children, medulloblastoma must be always excluded. The typical striated appearance can be absent in children.
Progressive nature of the symptoms	Helps to differentiate LDD from true dysplasia of the cerebellum.
Age, absence of cortical tubers, subependymal nodules and absence of white matter changes	Helps to exclude dysplasic cerebellar lesions from tuberous sclerosis
Age and absence of cystic tumours with an enhancing nodule	Helps to exclude cerebellar astrocytoma and haemangioblastoma. The former usually manifests in paediatric patients.
Age and absence of enhancement	Excluded medulloblastoma, which manifests in paediatric patients.
Age <50 years, solitary unilateral lesion, absence of intralesional haemorrhage and characteristic striated pattern	Excluded cerebellar metastases

BCVA, best-corrected visual acuity; LDD, Lhermitte-Duclos disease; y, years.

After LDD diagnosis, the physician should look for systemic signs of CS (box 1 and table 1), as CS is frequently associated to LDD and request the screening for a *PTEN* gene mutation (CC score ≥10).[20] This approach permitted us to diagnose CS in our patient. Management for CS is primarily focused on the cancer risks.[5–9] Tan *et al*[5] (2012) reported estimated lifetime risks of 85.2% for breast carcinoma, 35.2% for thyroid carcinoma, 28.2% for carcinoma of the endometrium, 9.0% for colorectal carcinoma, 33.6% for kidney cancer and 6% for melanoma.

Adults should undergo yearly thyroid ultrasound scan and dermatological evaluation. Beginning at age 30 years, women should undergo monthly breast self-examination; annual breast screening (mammogram, MRI) and transvaginal ultrasound scan or endometrial biopsy.[32] Beginning at age 35 years, colonoscopy should be performed in men and women with frequency dependent on degree of polyposis identified as well as biennial (every 2 years) renal imaging (CT or MRI preferred), beginning at age 40 years.[32] In those cases with a family history of a particular cancer type at an early age, the screening should be initiated 5–10 years prior to the youngest age of diagnosis in the family.[32]

The treatment for the benign and malignant manifestations of CS is the same as for their sporadic counterparts. Topical agents (5-fluorouracil), curettage, cryosurgery or laser ablation are rarely used to alleviate the mucocutaneous manifestations. The excision of cutaneous lesions is indicated if malignancy is suspected or if symptoms, deformity or scarring are present.[32]

In our patient, systemic screening excluded thyroid cancer recurrence as well as new malignancies of breast, genitourinary and gastrointestinal systems. The screening of systemic malignancy is fundamental in CS patients, as it can prolong life expectancy if cancer is detected early.[5–9]

LDD is a rare differential diagnosis of posterior fossa space occupying lesions it is a rare manifestation of the extremely rare CS.[2 30] Some clinical and imagiological findings (table 3) can help the physician to make the differential diagnosis. Our case supports the possibility of performing an early preoperative diagnosis of LDD based on pathognomonic MRI findings. OCT can be a useful tool for monitoring the oedematous status of the optic nerve after surgery. Funduscopy can be the starting point for the diagnosis of LDD and CS, leading to an adequate and prompt treatment of LDD and systemic screening for malignancies, which could be life-saving. Prompt treatment can even potentially prevent visual function loss, as occurred in our case.

This case should alert physicians to consider LDD in the differential diagnosis of a patient with papilloedema and a posterior fossa mass on neuroimaging.

Learning points

- Lhermitte-Duclos disease (LDD) is an extremely rare differential diagnosis of space-occupying lesions of the posterior fossa, but deserves to be considered.
- LDD is a slowly growing hamartoma of the cerebellum and has a pathognomonic appearance on MRI, a striated or tigroid pattern, permitting a preoperative diagnosis, which is supported by this case report.
- LDD is generally associated with Cowden syndrome, a rare phakomatosis associated with mutations in the *PTEN* gene and predisposing to multiple hamartomas and neoplasias.
- The prompt diagnosis and treatment of LDD can be life-saving and, in some cases, can prevent visual function loss. Surgery is indicated when intracranial hypertension develops. A ventricular shunt is placed initially, which is followed by tumour resection. Optical coherence tomography can be used to monitor the effect of surgery in reducing the oedematous status of the optic nerve.
- After LDD diagnosis, a systemic screening for Cowden syndrome manifestations must be performed to exclude this syndrome and the related malignancies, which can also be life-saving.

Contributors IFG and LDA have been involved in patient care. IFG was responsible for the review of clinical records of the patient and for manuscript drafting. LDA was responsible for the review of the manuscript and the final approval of the manuscript to be submitted for publication.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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