Six adult patients with septo-optic dysplasia and drug-resistant epilepsy: Clinical findings and course

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A B S T R A C T
Septo-optic dysplasia (SOD) is a rare disorder associated with optic nerve hypoplasia, pituitary abnormalities and agenesis/dysgenesis of midline brain structures including the septum pellucidum and corpus callosum. Though sometimes associated with drug-resistant epilepsy, this association has not been well studied. We report six SOD patients with associated malformation of cortical development (MCD) and drug-resistant epilepsy who underwent video-EEG telemetry at our centre between 1998 and 2016 for drug-resistant epilepsy. Three then underwent surgery; right temporal neocortical resection, right functional hemispherectomy and placement of a vagus nerve stimulator. Clinical findings and the patients' ultimate courses are discussed.

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1. Introduction
Septo-optic dysplasia (SOD) is an uncommon developmental anomaly, initially described in 1941 [1] and again in 1956 [2], that has an estimated incidence of roughly one per 10,000 live births [3]. Occurring in both males and females, it is classically characterized by the triad of optic nerve hypoplasia; midline brain abnormalities that include the absence of the corpus callosum and septum pellucidum; and hypothalamic-pituitary endocrine deficiencies [4–6]. However, it also appears in conjunction with a wide variety of cerebral anomalies, albeit most consistently with schizencephaly [7–13], such that the term SOD-Plus was recently proposed to describe SOD associated with cortical dysplasia [8]. The clinical presentation of SOD can range from mild to no symptoms [14,15] to severe developmental delay and incompatibility with life. It also can be associated with mild to severe visual impairment, sensory/neuronal hearing loss, and a range of other symptoms and signs that include a variety of endocrine disorders including precocious puberty, dwarfism and diabetes insipidus; other skeletal abnormalities; anosmia; and a range of cardiac anomalies, among others [5,7,8,10,16–22]. Seizures have also been described, which range from infantile spasms [23–25] to a variety of drug-resistant epilepsies presenting either during childhood or adulthood [4,5,9,11,13,19,20,26–31]. However, most published reports on patients with seizures and SOD are limited to a single case report or at most series that include a few patients. (See Table.)

In this report, we review the clinical features associated with SOD in six adult epilepsy patients. Ages ranged from 18-58 years. All had drug-resistant seizures and were seen between 1998 and 2016 for video-EEG telemetry in the Epilepsy Monitoring Unit (EMU) at the University Hospital at the London Health Sciences Centre (LHSC). These cases are first summarized individually and then analyzed collectively.
Case 1: A 57-year-old female and presented to the EMU with intractable seizures. Neuro-ophthalmic examination revealed absent visual fixation, pendular nystagmus and bilateral disc pallor, as well as fundoscopically-con firmed hypoplasia of both optic discs with visual impairment from age 6 months. Magnetic resonance imaging (MRI) of the brain revealed SOD, as well as open-lipped schizencephaly affecting both frontal lobes and pachygyria (Fig. 1a). Video-EEG monitoring revealed both epileptic and non-epileptic seizures, with the interictal EEG showing abundant multifocal slow waves “from several different areas of the brain”, and left temporal spikes during sleep. Thirteen non-epileptic spells were captured, characterized by either the sensation of shaking, right-sided weakness, and irregular shaking and grunting, all associated with preserved alpha rhythms and semiology of non-epileptic behavioral events. Five epileptic seizures occurred. These were characterized by generalization stiffening followed by asymmetric right arm tonic posturing, followed by symmetric tonic then clonic seizure activity with diffuse muscle and movement artifact, the onset non-localizable on EEG. However, the interictal EEG revealed abundant multifocal slow waves and left temporal spikes during sleep (Fig. 1b). The patient’s seizures were resistant to three different antiseizure medications.

Case 2: A 33-year-old male presented to the EMU with drug-resistant seizures. In addition to SOD, MRI of the brain showed bilateral perisylvian cortical dysplasia and polymicrogyria involving the frontal and parietal lobes, in addition to right-sided band heterotopia. (Fig. 2a). Prolonged EEG recording displayed independent bi-temporal and parietal lobes, in addition to right-sided band heterotopia, perisylvian cortical dysplasia and polymicrogyria involving the frontal hemispheres. In addition to SOD, MRI of the brain showed bilateral perisylvian cortical dysplasia and polymicrogyria, heterotopic grey matter and agenesis of corpus callosum. At that time, she underwent partial agenesis of the corpus callosum, hypogenesis of the anterior commissure and the optic chiasm, heterotopia and white matter changes.

2. Cases

| Case | MRI brain                                                                 | Hypothalamic-pituitary axis/MRI | Seizures       | Surgery            | Seizure outcome         |
|------|----------------------------------------------------------------------------|--------------------------------|----------------|--------------------|-------------------------|
| 1    | Bilateral frontal lobe schizencephaly and pachygyria                      | Normal                         | Normal Refractory | No Refractory      | Unchanged on ASDs       |
| 2    | Bilateral perisylvian cortical dysplasia and polymicrogyria               | Normal                         | Normal Refractory | VNS Refractory     | Unchanged on ASDs       |
| 3    | Multiple congenital abnormalities including cortical dysplasia, heterotopic grey matter and agenesis of corpus callosum. | Normal | Refractory Right functional hemispherectomy | Refractory Right temporal neocortical resection | Unchanged on ASDs       |
| 4    | Multiple congenital abnormalities including cortical dysplasia, bilateral closed lip schizencephaly, cortical heterotopia, absence of the septum pellucidum, small optic nerves bilaterally, a small optic chiasm and hypoplastic pituitary gland. | Normal | Refractory Right functional hemispherectomy | Refractory Right temporal neocortical resection | Unchanged on ASDs       |
| 5    | Multiple congenital abnormalities including, absence of the septum pellucidum, hypoplasia of chiasm and optic nerves, agenesis of corpus callosum and heterotopia. | Normal | Refractory No | Refractory No | Unchanged on ASDs       |
| 6    | Partial agenesis of the corpus callosum, hypogenesis of the anterior commissure and the optic chiasm, heterotopia and white matter changes. | Normal | Refractory No | Refractory No | Unchanged on ASDs       |

Figure 1A & B: A: Axial FSE T2 and Coronal FLAIR images: In the frontal lobes, bilaterally there is open-lipped schizencephaly. The lip extends anterior from the precentral gyrus into the lateral ventricles bilaterally. No heterotopia was present. Pachygyria is noted anterior and posterior to the clefts bilaterally. b: EEG of the patient in Case 1 with bipolar montage demonstrates: The posterior background activity contains a 11-Hz alpha rhythm. Insomnioc medium to medium-high amplitude 5-6 Hz bitemporal and intermittent bursts of theta were present. Occasional semi-rhythmic 2 Hz bifrontal predominant diffuse 1-5 second runs of delta were present. A-Spikes: Principally during drowsiness and wakefulness, broad involving the F7-M1-T3 derivations. C-Seizure: Sudden muscle artifact, then rhythmic 3 Hz, later obscured by muscle artifact ending abruptly approximately 83 s after electrogaphic onset. Clinical onseet preceded electrogaphic onset. Reword: Semiology involved the patient grabbing both knees first and then the hand rails, prior to elevating his right arm/ flexed at the elbow prior to evolution to a generalized motor seizure with post-ictal somnolence. Patient grabs knees with hands, holds hand rail, then elevates right arm, flexed at elbow, then tonically slumps off bed with a secondary generalized seizure; sleeps postictally.

Figure 2A: Axial FSE T2 and Coronal FLAIR images: In the frontal lobes, bilaterally there is open-lipped schizencephaly. The lip extends anterior from the precentral gyrus into the lateral ventricles bilaterally. No heterotopia was present. Pachygyria is noted anterior and posterior to the clefts bilaterally.
Figure 1A & B (continued).
her seizures. In 2000, a complete right hemispherectomy was performed, after which she was seizure-free for seven years, until 2007 when seizures returned. In 2008, EEG revealed spikes in the left frontal central and temporal region, and multiple independent spikes. She also exhibited continuous epileptiform discharges, consisting of a mixture of polyspike- and slow spike-and-waves in the left frontal and centrotemporal region, as well as abundant multifocal epileptiform discharges independently in the same region and elsewhere. There also was evidence of slow background activity and diffuse intermixed slowing (Fig. 3a). Post-operative MRI demonstrated a right post-operative changes with an area of gliosis surrounding the surgical site (Fig. 3b). Given the presence of left hemispheric spikes and spike-and-waves and waves suggesting the potential for independent left hemispheric seizures, she was treated medically and currently remains on multiple antiseizure medications.

Case 4: A 29-year-old female presented with drug-resistant seizures, as well as a history of partial anterior pituitary insufficiency and infertility, for which she was on hormonal replacement. Physical examination revealed prominent eye abnormalities, congenital nystagmus (left > right), a partial left third nerve palsy with exotropia, convergence insufficiency of the left eye, and small, hypoplastic and atrophic optic discs bilaterally (left > right) (Fig. 4a).

MRI of the brain demonstrated bilateral closed lip schizencephaly with an abnormally thickened insular and opercular cortex bilaterally, likely representing a neuronal migration abnormality. There also was absence of the septum pellucidum, hypoplastic optic nerves bilaterally, a small optic chiasm, and a hypoplastic pituitary gland (Fig. 4b). On EEG, seizures arose from the right temporal lobe, but there also were multifocal interictal epileptiform discharges present. Sleep activated generalized poly-spikes that were maximal in the right temporal region, with enhanced activation in the right mid-temporal-central region and generalized spikes and spike-and-waves and waves (Fig. 4c). She underwent a right temporal neocortical resection in 2006, after which the frequency of her seizures declined by roughly 50%.

Case 5: A 37-year-old male presented with drug-resistant seizures and intellectual disability. MRI of the brain revealed callosal dysgenesis with small nodular densities in the periventricular white matter of both frontal lobes, consistent with heterotopias. The optic chiasm and optic nerves were reduced in size. No septum pellucidum could be identified (Fig. 5a). On EEG, there were multifocal epileptiform discharges (Fig. 5b).

Case 6: A 58-year-old male with longstanding epilepsy since childhood was referred for drug-resistant epilepsy and newly diagnosed septo-optic dysplasia. His medical history remarkable for non-alcoholic steatohepatitis (NASH); esophageal varices, secondary to his liver disease (which had required banding); which had required banding; chronic bilateral leg lymphedema; obstructive sleep apnea, for which he had self-discontinued CPAP; type II diabetes mellitus; and essential tremors.

Brain MRI demonstrated SOD with partial agenesis of the corpus callosum. Focal tiny subependymal heterotopias were also identified along the margins of the right lateral ventricle. There was hypogenesis of the anterior commissure and the optic chiasm. Aside from this, there was scattered high T2 signal focus in the white matter of both hemispheres, which was believed to indicate micro-angiopathic changes (Fig. 6a). EEG telemetry recorded several non-epileptic events in addition to a possible focal seizure, associated with insufficient cortical representation and no other EEG abnormalities (i.e., a normal wakeful and sleep EEG).

3. Discussion

Septo-optic dysplasia refers to a heterogeneous group of disorders that invariably include optic nerve and/or optic chiasma hypoplasia, accompanied by either the absence or dysgenesis of the septum pellucidum [4–6], as well as hypothalamic-pituitary insufficiency. SOD-Plus, which exhibits the classic triad of findings in addition to other evidence of cortical malformation, is currently believed to primarily be a genetic disorder that affects multiple stages of cortical

![Fig. 2A](image-url) A-D demonstrating brain MRI with spin echo sequences. Spin echo shows R fast Perisylvian cortical dysplasia involving the left frontal lobe and parietal lobe opercula and, on the right, primarily the frontal lobe. Band heterotopia is also evident in the right corona radiata white matter. EEG displayed in a bipolar montage with background: 8–9 Hz, medium voltage, symmetric and reactive. spikes were seen at F8-T4-A2-Fp2 during wakefulness and sleep. Theta: Polymorphic 5–6 Hz, medium voltage, posterior temporal in both hemisphere, as well as a small amount of generalised activity.
development, as opposed to being caused by an acquired etiology—like infection, infarction or hemorrhage—often implicated in isolated SOD [32–34] or schizencephaly alone [35–37]. Indeed, only a small percentage of isolated SOD cases are believed to have a predominantly genetic basis [35,38]. Seizures are not uncommon in these patients, though most reports describe only a single or at most a few cases [4,5,9,11,13,19,20,23–31]. In perhaps the only relatively large series, from Spain, 11 of 20 patients with documented SOD (55%) also had documented epileptic seizures [28]. Meanwhile, in a series of 100 patients with optic nerve hypoplasia, 12% had documented seizures; however, only 10% of the 100 had confirmed SOD, and how many of these 10 had seizures was not reported [19].

In our series of six patients with MRI-documented SOD and EEG-documented seizures, there were three males and three females. The age at presentation to our EMU ranged from 18 to 58 years (mean 38.7, standard deviation 15.9). Ophthalmological studies, MRI, and dynamic pituitary function tests make the Diagnosis of SOD in our patients without genetics testing.

The International League Against Epilepsy (ILAE) has proposed the following definition of drug-resistant epilepsy and suggests that this term be used instead of the term ‘refractory epilepsy’ [39]. Drug-resistant epilepsy occurs when a person has failed to become (and stay) seizure free with adequate trials of two seizure medications. By this definition, all of our patients with SOD were referred to the epilepsy clinic for drug-resistant epilepsy. In addition, consistent with the heterogeneous ocular manifestations of SOD [2,17,38], three of the six had visual impairment. In addition, two of our patients had combined hypothalamic–pituitary disturbance, with growth hormone (GH) deficiency the most common pituitary endocrinopathy (33.3%), followed by hypothyroidism and adrenal insufficiency (16.6%); one female also suffered from infertility which may also have been endocrine-related. Only a single patient had multiple pituitary hormonal deficiencies. One patient had global developmental delay and three had intellectual and/or behavioral deficits; overall, mild to moderate neurological deficits were observed in four of the six patients.

On MRI, bilateral optic nerve hypoplasia was identified in 50% of the patients. Anatomical brain MRI also revealed very heterogeneous morphological anomalies, ranging from isolated agenesis of the septum pellucidum in one case, to multiple malformations involving the cerebral cortex, with some malformation in cortical development identifiable in five out of the six patients. These malformations
included schizencephaly, pachygyria, polymicrogyria, cortical dysplasia and heterotopias. On video-EEG monitoring a range of different seizure types were identified, and seizures had a multifocal origin in four patients. In five of the six patients, a malformation of cortical development contributed directly to drug resistant epilepsy.

Focal cortical dysplasia (FCD), a malformation of cortical development, is the most common non-tumor cause of drug-resistant epilepsy in the pediatric population and the second or third most common etiology of drug-resistant seizures in adults [37,40–44]. It also is among the abnormalities most commonly associated with schizencephaly, which may appear either unilaterally or on both sides of the brain [9]. The clinical presentation of schizencephaly generally depends upon the quantity and location of brain involvement.

The rate and degree of success surgically controlling seizures in patients with focal cortical dysplasia, schizencephaly and other cortical malformations largely depends on the number of seizure foci and their proximity to each other and, hence, resectability [45]. Some form of surgical intervention was performed in three of our six patients, but none of the patients became seizure free. Functional hemispherectomy was performed in one patient, and this resulted in greater than a 50% reduction in seizure frequency. Similarly, in a second patient, right temporal neocortical resection was done, and this resulted in about a 60% reduction in seizure frequency. In a third patient, implantation of a vagus nerve stimulator (VNS) was considered less effective, reducing seizure frequency by <50%. The three remaining patients were not considered surgical candidates and currently remain on multiple antiseizure medications for ongoing management. This being said, one of these three patients had previously been seizure free for seven years following a complete right hemispherectomy at age 20 for strictly right-sided seizures. When we saw her, however, she exhibited diffuse epileptiform discharges, so that no further surgery was recommended. Still, respective epilepsy surgery has been effective in seizure control for selected patients with a congenital or early-acquired brain lesion and drug-resistant encephalopathic generalized

![A) B](image)

**Fig. 3.** a: Background: 5–6 Hz, low voltage, poorly-developed and not well-sustained. Theta: Low frequency 4–5 Hz, low-medium voltage, diffuse, persistent, maximal in the left hemisphere. Delta: 2 Hz, low voltage, arrhythmic, left frontal - central - temporal and generalised, persistent, more prominent in amplitude and persistent on the left side. Spikes and Waves: Excessive, nearly-continuous slow spikes and waves (approximately 1.5–2.5 Hz) in the left hemisphere. b: A, Axial FLAIR. B&C Axial FSE T2. High T2 signal demonstrated on FLAIR images surrounding the area of encephalomalacia is most consistent with post-surgical gliosis. Right subtotal hemispherectomy with areas of gliosis surrounding the surgery site. Multiple congenital abnormalities including cortical dysplasia in the anteromedial right frontal lobe, heterotopic grey matter and agenesis of the corpus callosum. There is significant dilatation of the occipital horn of the left ventricle. There are severe abnormalities involving the grey matter bilaterally. There is severe cortical dysplasia of the left temporal and occipital lobe and heterotopic subependymal grey matter surrounding the anterior and posterior horn of the left lateral ventricle, in addition to bilateral frontal cortical dysplasia.
epilepsies such as Lennox-Gastaut syndrome (LGS) despite diffuse or multifocal EEG finding [46].

4. Conclusions

Septo-optic dysplasia is a syndrome that varies tremendously in its presentation, sometimes occurring in isolation and sometimes in association with other cortical malformations. In six patients with SOD plus who presented to our epilepsy clinic with drug-resistant epilepsy, cortical malformations commonly were implicated as the etiology for epilepsy. Seizures tended to be multifocal and at best only partially responsive to seizure surgery, when amenable to surgery at all.

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Fig. 4. A: Left hypoplasia of the optic discs with double margins. B: Left optic disc pallor. a: AX STEREO ZIPPED, B&c AXIAL T2. D Sagittal PLANE LOC: Bilateral closed- lip schizencephaly with abnormally-thickened insular and opercular cortex bilaterally, likely representing a neuronal migration abnormality like cortical heterotopia. There is also the absence of the septum pellucidum with small optic nerves bilaterally, a small optic chiasm and a hypoplastic pituitary gland. c4 Bipolar montage: Posterior background rhythm 8 Hz. A/Left frontocentral and right temporal spikes. Seizure onset over right temporal lobe. D/Sleep: Activated generalized polypsikes (max right temporal). Enhanced activation in right mid temporocentral region and generalized spikes and spikes and waves.
Fig. 4 (continued).
Fig. 5. a: coronal FLAIR. B and C: Axial FSE T2 Callosal dysgenesis with small nodular densities in the periventricular white matter of both the right and left frontal lobes consistent with heterotopia. The optic chiasm and optic nerves are somewhat small. No septum pellucidum is identified. b A: Bipolar montage, spikes were seen at F4-F3 during wakefulness and sleep. B: spikes were seen at F7-T3-T5.
Fig. 5 (continued).
Fig. 6. a, B, C T1 Axial showing hypoplasia of the optic nerves, optic chiasm and optic tracts bilaterally in addition to a small pituitary gland.