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Case Report

CASE REPORT

Parry Romberg syndrome presenting with a giant intracranial aneurysm: a case report

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Abstract

A giant intracranial aneurysm was diagnosed in a 10-year-old girl when she developed a right abducens nerve palsy. The aneurysm was treated successfully. Six years later, however, she presented with a progressive *en coup de sabre* deformity, leading to the diagnosis Parry Romberg Syndrome (PRS), a rare diagnosis characterized by hemifacial atrophy of skin, subcutaneous tissue, skeletal muscle and bones and often associated with various non-specific intracerebral abnormalities. In this patient retrospective analysis of computed tomography and magnetic resonance imaging imaging indeed revealed intracerebral calcifications and aspecific white matter lesions. Remarkably, in this patient the giant intracranial aneurysm turned out to be the presenting symptom of PRS years before the characteristic facial deformities became apparent.

INTRODUCTION

Parry Romberg Syndrome (PRS) is a slowly progressive syndrome characterized by hemifacial atrophy of skin, subcutaneous tissue, skeletal muscle and bones. It is a heterogeneous disorder with an incompletely understood pathogenesis. Several etiologies have been proposed, including auto-immunity, vascular dysfunction, trauma, infection, autonomic dysfunction, neural crest migration disruption and a possible genetic predisposition [1, 2]. PRS is suggested to be part of a larger disease entity, which encloses linear scleroderma en coup de sabre as well [1, 3].

The most frequent systemic manifestations of PRS are neurologic, affecting up to 20% of patients. These neurological symptoms include seizures, migraines (sometimes leading to hemiplegia), brain atrophy, cranial neuropathies, intracranial aneurysms and other intracranial vascular abnormalities [1, 2, 4]. Ophthalmologic abnormalities include enophthalmos, uveitis, retinal vasculitis, ptosis and abnormal ocular movements due to muscle paresis or weakness [1, 2, 5]. Dermatologic, cardiologic, auto-immune, endocrinologic and infectious manifestations have also been described [2]. There seems to be a female predominance in patients with PRS [6].

Radiological abnormalities on computed tomography (CT) and magnetic resonance imaging (MRI) in PRS patients include aspecific white matter lesions, mild cortical thickening, intracerebral calcifications and/or atrophy and leptomeningeal enhancement [2].

CASE REPORT

We report a 10-year-old girl who presented with symptoms of a right abducens nerve palsy. Except for intermittent headaches,

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occurring approximately once every month, there were no other clinical symptoms. Neurological examination showed no other abnormalities, including normal ophthalmoscopy. A small intradermal capillary hemangioma was observed on the right upper eyelid.

CT and MRI of the brain showed a giant intracranial aneurysm located at the siphon of the internal carotid artery, about 2.3 cm in diameter, as the cause of the abducens nerve palsy (Figs 1 and 2). She was referred for neurosurgery, where balloon occlusion of the aneurysm was performed successfully.

Postoperatively, we observed minimal right sided ptosis and slight difference in pupil size, the right pupil being smaller (Fig. 5). The patient complained of headaches along the first branch of the trigeminal nerve, which was successfully treated with carbamazepine for 6 months.

On routine check-ups, a progressive en coup de sabre deformity was noted on the right forehead, where originally the hemangioma was located (Figs 6–8). Retrospectively, the brain CT showed right occipital intracerebral calcifications (Fig. 3). Furthermore, the brain MRI showed white matter lesions in the area of these intracerebral calcifications, possible vascular degenerative (developmental venous angioma) (Fig. 4). The patient still complained of right sided migraines. Based on these findings, the diagnosis of PRS was made 6 years after surgery.

Currently, the patient is 20 years old. Routine yearly check-ups including brain imaging show a slight progression of the hemifacial atrophy, unchanged appearance of the occluded aneurysm and no expansion of intracranial calcifications and white matter lesions (Fig. 9). Furthermore, no additional aneurysms have been found.

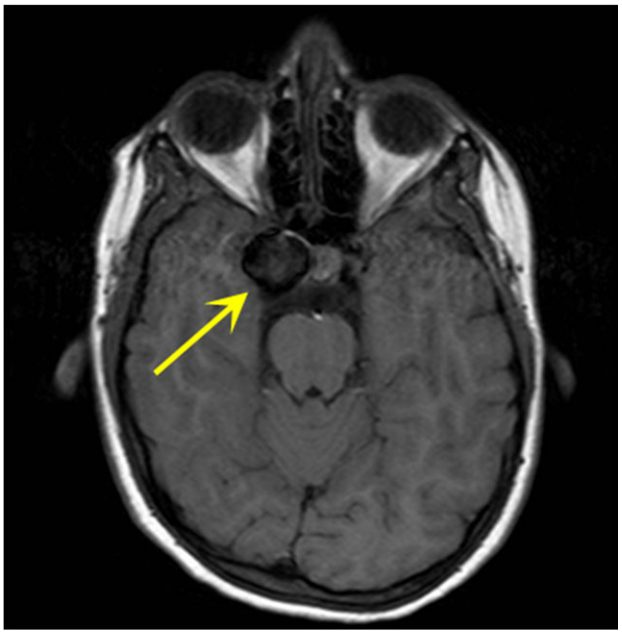

Figure 1: Giant intracranial aneurysm at the siphon of the internal carotid artery.

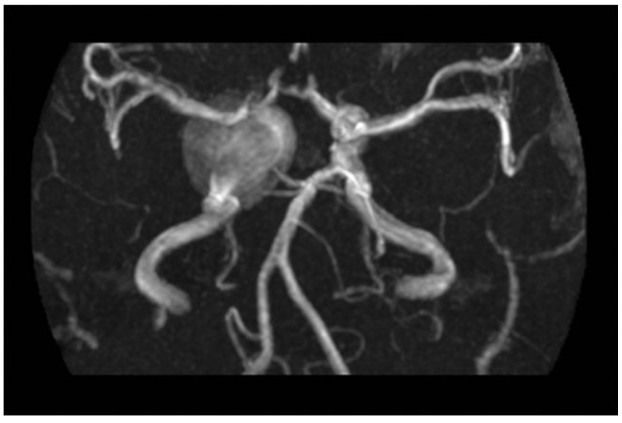

Figure 2: Magnetic resonance angiography at presentation showing the giant intracranial aneurysm and onset ectasia of the left carotid siphon.

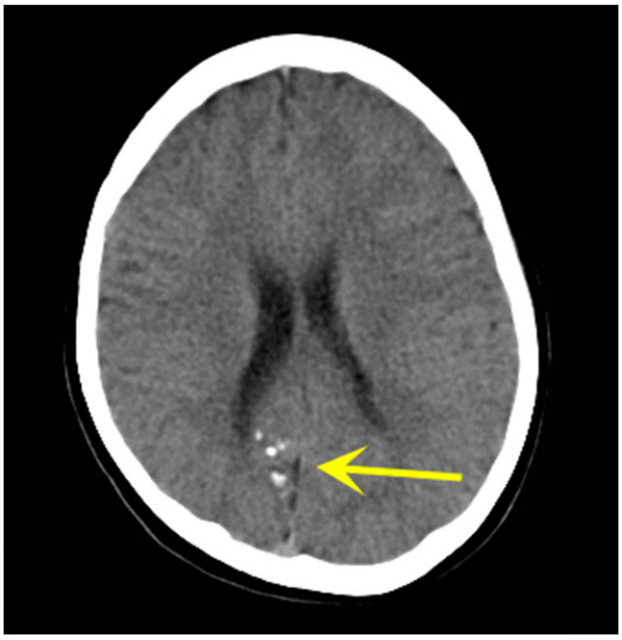

Figure 3: Right occipital intracerebral calcifications on preoperative CT.

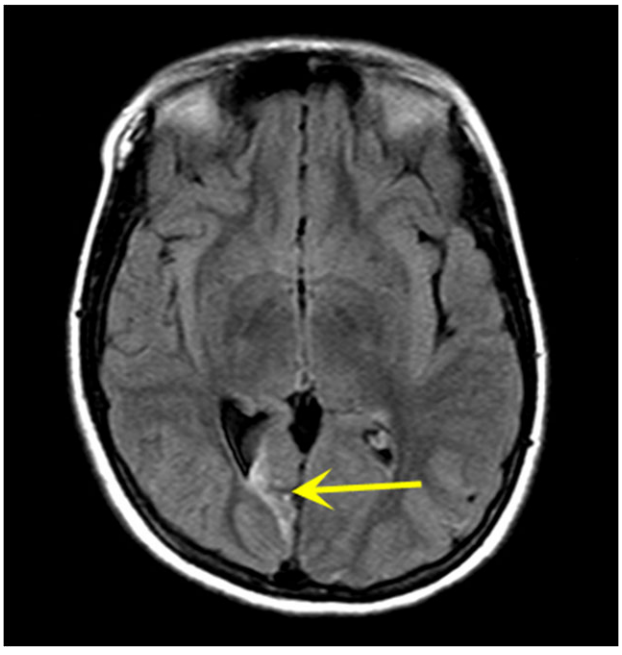

Figure 4: Preoperative MRI showing white matter lesion in area of CT calcifications, possible vascular degenerative (developmental venous angioma).

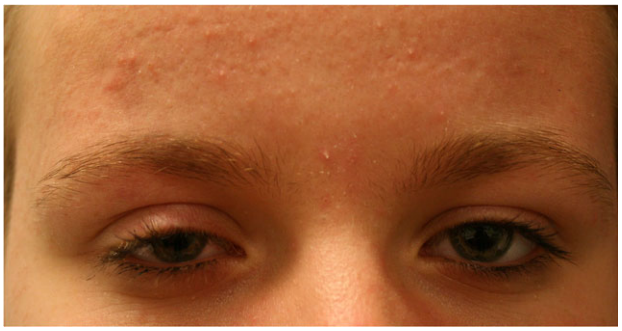

Figure 5: One and a half years after surgery. There is ptosis and miosis of the right eye. Note the intradermal capillary hemangioma on the upper eyelid of the right eye.

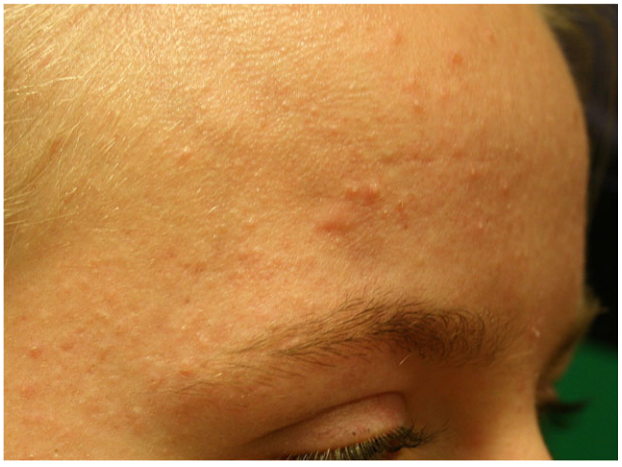

Figure 6: One and a half years after surgery. Beginning of hemifacial atrophy on the right forehead.

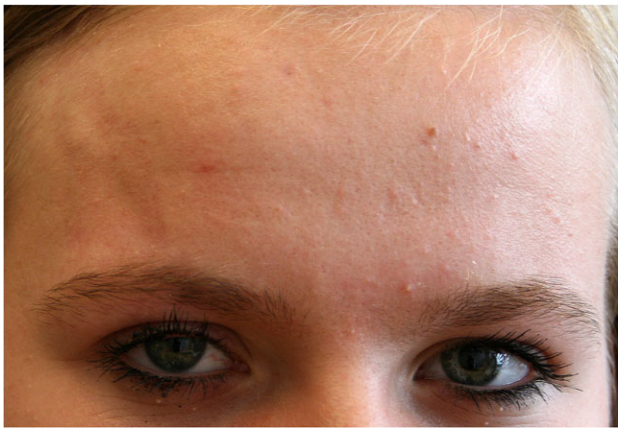

Figure 7: Three and a half years after surgery. Progressive hemifacial atrophy.

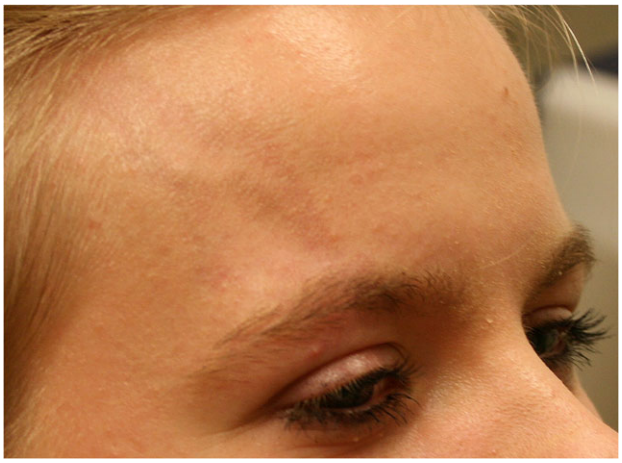

Figure 8: Three and a half years after surgery. Progressive hemifacial atrophy.

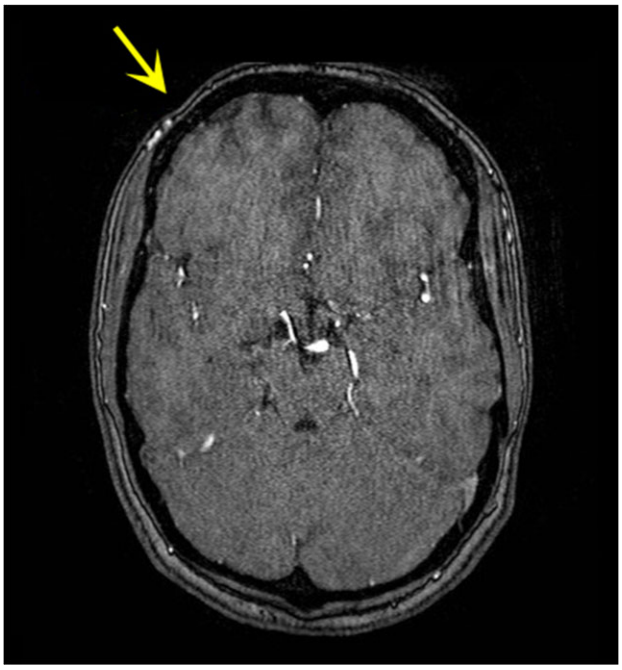

Figure 9: MRI 10 years after surgery showing right sided frontal subcutis atrophy.

DISCUSSION

PRS is a rare syndrome characterized by hemifacial atrophy of skin, subcutaneous tissue, skeletal muscle and bones. While symptoms are mostly neurological, various other organ systems can be affected [2, 4–6].

Intracranial aneurysms in PRS are rarely reported [7–10]. In these cases, the patients were already diagnosed with PRS years before aneurysm development. By contrast, in our patient, the initial symptom was a right abducens nerve palsy caused by an intracranial aneurysm of the internal carotid artery. The diagnosis PRS was only made a few years later, when the hemifacial atrophy became more apparent. Both the hemifacial atrophy and the intradermal capillary hemangioma were located on the right forehead, but the location of the hemangioma did not fully correspond with the extent of the atrophy. Therefore, the atrophy cannot be explained by the presence of the hemangioma.

To our knowledge, this is the third report describing an association between PRS and intracranial aneurysms in childhood [8, 10]. Bosman *et al.* [8] describe a 10-year-old patient with a giant intracranial aneurysm and two additional smaller intracranial aneurysms. Schievink *et al.* [10] report a patient with PRS who

had multiple and progressive intracranial aneurysms that required treatment at the age of five, 12 and 21 years old.

In conclusion, there might be an association between PRS and intracranial aneurysms and a intracranial aneurysm can be the presenting symptom of PRS. In case of a intracranial aneurysm in PRS, these aneurysms can be multiple and progressive, therefore regular follow up is advised.

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We thank our patient for her consent to publish the case and the related pictures.

CONFLICT OF INTEREST STATEMENT

No conflicts of interest.

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ETHICAL APPROVAL

Informed consent was taken from the patient for publication of the case report.

CONSENT

Patient's written consent was taken for publication.

GUARANTOR

L.A. Bok will act as guarantor.

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