

Rare disease

Aneurysmal subarachnoid haemorrhage in Parry–Rhomberg syndrome

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Summary

Parry–Romberg syndrome (PRS) or progressive hemi facial atrophy syndrome is a rare condition of unknown aetiology that is characterised by progressive unilateral facial and cranial atrophic changes of skin, subcutaneous tissues and bone. The authors describe a 37-year-old female with a history of PRS, who presented with a subarachnoid haemorrhage secondary to rupture of a 9 mm fusiform aneurysm of the posterior cerebral artery. There was an associated external carotid arterio-venous fistula noted with this aneurysm. The aneurysm was treated by endovascular route and was successfully coiled. Follow-up angiogram revealed spontaneous resolution of the fistula with good occlusion of the aneurysm. The aetio-pathogenesis of this rare occurrence, literature review and its management is discussed.

BACKGROUND

Parry–Rhomberg syndrome (PRS) is a rare condition and associated intracranial vascular abnormality is a rare occurrence. In our case, we describe the first case of posterior communicating artery aneurysm causing subarachnoid haemorrhage. This was incidentally associated with an external carotid arterio-venous (AV) fistula. The aneurysm was successfully managed by coiling and subsequently the AV fistula regressed spontaneously. We have written this case up due to the rarity and we also describe the aetio-pathogenesis of this condition with review of literature

CASE PRESENTATION

A 37-year-old lady with a background of PRS, presented with a history of sudden onset severe occipital headache associated with a right hemiparesis. Her Glasgow coma scale at admission was 13/15 and had a right-sided hemianopia. A non-contrast CT showed a diffuse subarachnoid haemorrhage with intraventricular extension (figure 1). Cerebral angiogram demonstrated a left P2/P3 posterior cerebral artery (PCA) aneurysm (figure 2) associated with a left external carotid artery AV fistula (figure 3). Significant dysplastic collaterals supplying the left anterior cerebral artery territory were seen arising from the right internal carotid artery (ICA). In addition collateralisation was seen from the left external artery through the middle meningeal supplying the left parietal cortex. These findings were noted in the previous angiogram performed 3 years prior to this presentation.

She presented 3 years ago, when she was 32 weeks pregnant, with left-sided facial swelling associated with a throbbing sensation, left ear pulsatile tinnitus, jaw pain and left facial erythema. These symptoms gradually improved over the subsequent week. An MRI brain at that stage showed a PCA aneurysm and left hypoplastic ICA. Subsequent six-

vessel angiogram confirmed an 8 mm fusiform aneurysmal dilatation of the left P2/P3 segment of the posterior communicating artery. She was treated conservatively owing to her clinical improvement. In her current presentation, the PCA aneurysm was noted to be 9 mm in size.

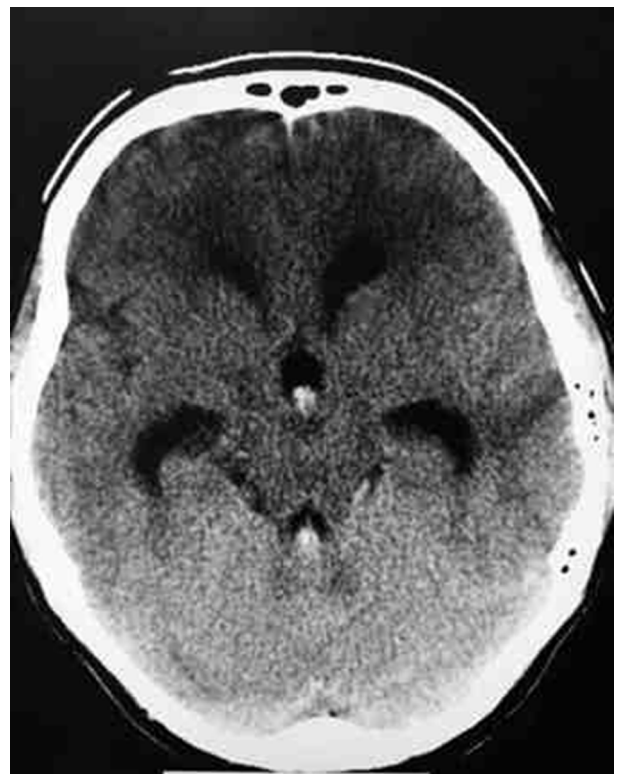


Figure 1 CT non-contrast brain demonstrating subarachnoid haemorrhage.

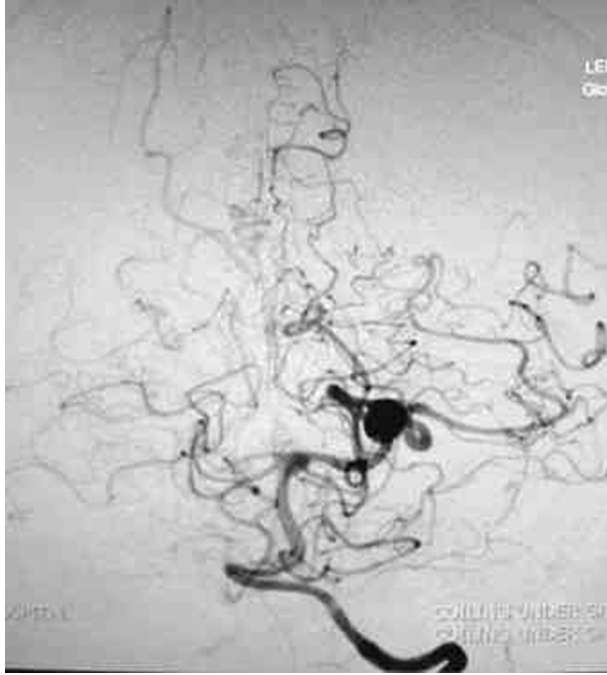


Figure 2 Cerebral angiogram demonstrating 9 mm P2/P3 PCA aneurysm.

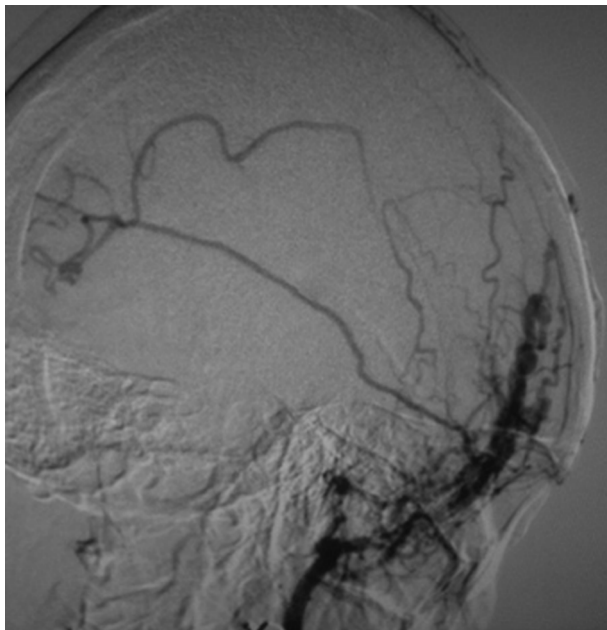


Figure 3 Angiogram demonstrating the dural AV fistula.

DIFFERENTIAL DIAGNOSIS

- ▶ AV malformation
- ▶ Aneurysmal subarachnoid haemorrhage.

TREATMENT

After discussing the treatment options, she was considered for endovascular management. Wada test was performed, which resulted in slight facial weakness and mild dysphasia, prior to the embolisation. She made an uncomplicated



Figure 4 Post coiling angiogram with complete occlusion of the aneurysm.

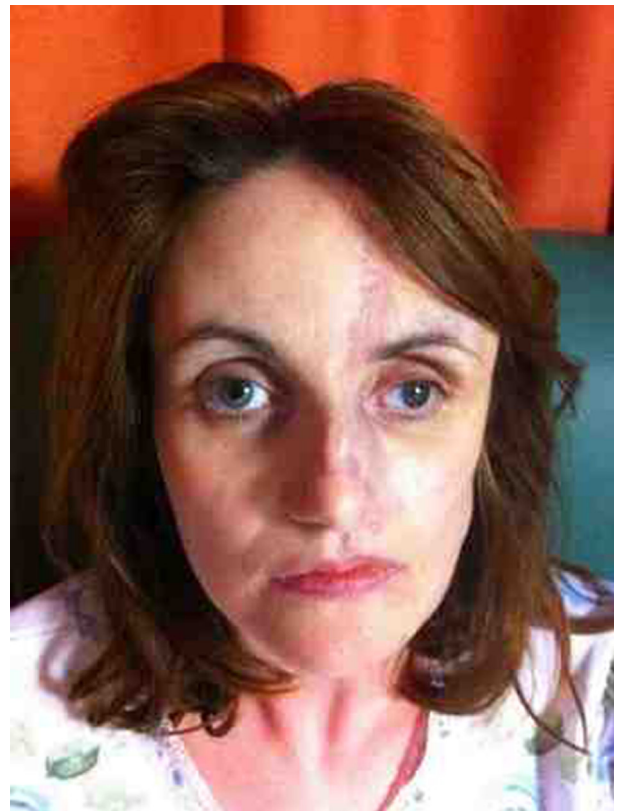


Figure 5 Facial appearance of Parry–Rhomb syndrome (eyes morphed).

postoperative recovery and the aneurysm was completely occluded (figure 4). CT brain post coiling revealed a small left PCA infarction with no neurological sequelae. Interestingly, subsequent angiogram at 15 months post coiling showed resolution of the left external carotid AV fistula.

Table 1 Summary of publications on intracranial aneurysms in PRS

Year	Author	Age	Location	Type	Management
1996	Schievink <i>et al</i> ⁶	5	Ipsilateral ICA	Giant fusiform	Carotid artery ligation and bypass
		12	Contralateral ICA	Giant fusiform	Carotid artery ligation and bypass endovascular
		21	PCA	Saccular	Endovascular
			Distal PCA	Fusiform	
2002	Pichiecchio <i>et al</i> ⁷	32	Ipsilateral MCA	Saccular	Conservative
2006	Aoki <i>et al</i> ⁴	56	Ipsilateral ICA	Giant Fusiform	Conservative
2009	Bosman <i>et al</i> ²	10	Ipsilateral ICA	Giant Fusiform	Ligation with high flow bypass
2011	Current case	35	Ipsilateral PCA	Fusiform	Endovascular embolism

ICA, internal carotid artery; MCA, middle cerebral artery; PCA, posterior cerebral artery.

OUTCOME AND FOLLOW-UP

She remains clinically well with a mild improvement in her right homonymous hemianopia.

DISCUSSION

PRS is a rare condition of unknown aetiology comprising of progressive atrophy of skin, subcutaneous tissue, fat and muscles of one side of the face. This is also known as progressive hemi facial atrophy. The onset is usually during the first or second decade progressing over a period of 2 to 10 years and then spontaneously remitting leaving the patient with considerable disfigurement. In individuals with this disorder, initial changes usually involve the tissues above maxilla or nasolabial fold and progress to involve the angle of mouth, eye, brow, ear and/or neck (figure 5).¹ Overlying skin may become either hyper or hypopigmented. Neurological sequelae include epilepsy, migraine and facial pain and brain lesions on CT and MRI such as ipsilateral cerebral atrophy or calcifications.¹ Intracranial vascular malformations including intracranial aneurysm have been rarely described in PRS.²⁻⁷ Table 1 summarises the available relevant literature on PRS and intracranial aneurysms.

Aetio-pathogenesis

Two current hypotheses on the aetiology of PRS present in the literature provide explanations as the origin of intracranial aneurysm in the disease. First it has been postulated that a failure of neural crest migration may explain some of the features of Perry-Romberg including vascular abnormalities and aneurysm formation.^{5,7,8} There are a number of features of vascular malformations that seem to support this hypothesis in the literature. First, most of the lesions are ipsilateral to the facial atrophy with a recent study on intracranial vascular malformations in PRS describing eight cases of intracranial vascular malformation including their own all reporting this feature.² Second, our own and two previous studies have described hypoplasia of the ICA in PRS.^{8,9} Congenital hypoplastic ICA is rare, and has been noted to be consistent with a failure of embryological development of the ICA, which forms by fusion of a succession of embryological arterial segments that lead from the carotid bifurcation in the neck to the Circle of Willis.¹⁰ Finally, there are common progenitors in the neural crest during development that derive the tunica media of cervicocephalic arteries and mesenchymal structures of the head and neck, such as the facial dermis, subcutaneous tissues, muscle and skeleton—all structures effected by Perry-Romberg syndrome.¹¹ How these factors could result in aneurysm formation remains unexplained, however it

seems plausible that the coexistence of aneurysm with other lesions of known developmental origin makes a strong case for a developmental abnormality in Parry-Romberg.

A second hypothesis for the aetiology of PRS postulates that it is caused by a localised vasculitis and it has been postulated that Parry-Romberg is a variant of linear scleroderma.¹² This is supported by histological examinations of facial specimens from patients with PRS that noted a proliferative interstitial neurovasculitis.¹³ In addition, there appears to be a propensity for giant ICA aneurysm, and fusiform aneurysms in case reports published in the literature (table 1).

These aneurysms are thought to arise from dissection of the arterial wall, and have been associated with both arterial wall inflammation (atherosclerosis) and connective tissue diseases such as Marfan’s and Osteogenesis Imperfecta, which result in underlying arterial wall weakness.¹⁴ Still, most cases of fusiform intracranial aneurysm are of unknown origin likely resulting from an unidentified vascular pathology.¹⁵ If an underlying inflammatory vasculitis was present this may explain the development of fusiform aneurysm, however, it would not account for the focal nature of the disease.

Learning points

- ▶ Subarachnoid haemorrhage caused by intracranial aneurysm rupture in Perry-Romberg syndrome is rare.
- ▶ Fusiform aneurysms in this clinical setting may pose a management challenge and the possibility of aneurysmal subarachnoid haemorrhage should be anticipated in this group of patients when intracranial aneurysms are detected.
- ▶ The cerebral aneurysm may be accompanied by other vascular malformation like AV fistula and these may not need treatment as demonstrated in our case.
- ▶ Endovascular treatment appears to be a safe option in the management of subarachnoid haemorrhage.
- ▶ Radiological follow-up by magnetic resonance angiography or cerebral angiogram may prove beneficial.

Competing interests None.

Patient consent Obtained.

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