

Ocular Abnormalities in Parry-Romberg Syndrome

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บทคัดย่อ

Parry-Romberg syndrome เป็นโรคที่พบได้ไม่บ่อย และมีลักษณะเฉพาะคือมีการฝ่ออย่างค่อยเป็นค่อยไปของผิวหนังและเนื้อเยื่อข้างใต้ รวมถึงกระดูกอ่อนและกระดูกบริเวณใบหน้าครึ่งหนึ่ง รายงานผู้ป่วยชายอายุ 65 ปี ซึ่งป่วยเป็นโรคนี้และมาพบแพทย์ด้วยการมองเห็นแคบลง ผลการตรวจตาพบความผิดปกติหลายอย่าง ประกอบไปด้วย การยุบของลูกตาเข้าไปในเบ้าตา กระจุกตาและรูม่านตาผิดปกติ ม่านตามีการฝ่อ มีความผิดปกติของเมดูลีในจอตา และการกลอกตาผิดปกติ ผู้ป่วยได้รับการตรวจเอ็กซเรย์พิเศษ และคลื่นไฟฟ้าทางตา และนำเสนอผลการตรวจในรายงานฉบับนี้ **จักขุเวชสาร 2554; มกราคม-มิถุนายน 25(1): 43-48.**

Original Article/นิพนธ์ต้นฉบับ

Ocular Abnormalities in Parry-Romberg Syndrome



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Abstract

Parry-Romberg syndrome is a rare disorder, characterized by progressive hemifacial atrophy involving the skin, soft tissues, cartilage and bone. The authors report a 65-year-old man with right hemifacial atrophy who presented with a complaint of visual field constriction. Multiple ocular abnormalities were detected including enophthalmos, corneal disorders, iris atrophy, pupillary abnormality, retinal pigment alteration, and restrictive strabismus. Neuro-imaging and ocular electrophysiology were performed and the results are presented. **Thai J Ophthalmol 2011; January-June 25(1): 43-48.**

Introduction

Parry-Romberg syndrome is a rare disorder, characterized by progressive unilateral wasting of facial skin and subcutaneous tissue with variable involvement of muscle, cartilage, and bone. The syndrome is usually a sporadic disease with onset in the first or second decade followed by a “burning out” of the atrophic process and subsequent stability.^{1,2} The etiology is still unknown. Sympathetic dysfunction, trauma, viral infection, autoimmune disorder and cranial vascular malformation are proposed causes.^{2,3}

Ophthalmologic disorders have been reported including enophthalmos, eyelid atrophy, iris abnormalities, uveitis, retina and optic nerve dysfunctions.¹⁻⁸ The authors report a case of Parry-Romberg syndrome who had multiple ocular abnormalities and presented with a constriction of the peripheral visual field.

Case report

A 65-year-old man presented with a complaint of painless decrease in the peripheral visual field in the right eye for 3 weeks. He had a history of slowly progressive atrophy of the right side of his face since the age of 10. Physical examination revealed generalized atrophy of subcutaneous tissue and muscles over the right half of the face with ipsilateral enophthalmos (Figure 1). There was no tongue or pharyngeal muscles involvement and there was no other neurological deficit.

Ophthalmological examination revealed atrophy of the right upper and lower eyelid with loss of right eyelashes and eyebrow. He had 6-mm right enophthalmos. His best corrected visual acuity was 20/80 in the right eye and 20/20 in the left eye. Slit lamp examination revealed peripheral corneal degeneration on the right upper cornea. There were inferior, fine keratic precipitates but no anterior chamber reaction. There was dense nuclear sclerosis, genera-

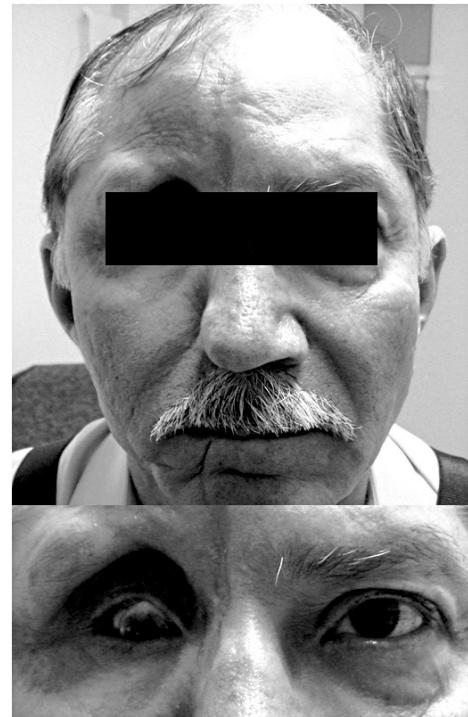


Figure 1 Patient's photographs show right hemifacial atrophy, right enophthalmos and loss of eyelashes and eyebrow. There is a paramedian forehead scar “en coup de sabre” separating atrophic from normal tissue. (รูปถ่ายผู้ป่วย)

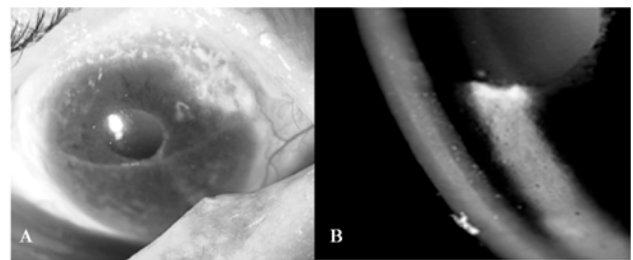


Figure 2 Right anterior segment photographs show peripheral corneal degeneration, iris atrophy, loss of pupillary ruff (A) and keratic precipitates (B). (รูปถ่ายผู้ป่วย)

lized iris atrophy and loss of pupillary ruff in the right eye (Figure 2). The right pupil was 3 mm and sluggishly reactive to light. The left pupil was 5 mm and normoreactive to light. There was no afferent pupillary defect. Ophthalmoscopy revealed a large area of generalized retinal pigment epithelium (RPE) atrophy in the lower half of the right retina (Figure 3). Left fundus examination was normal.

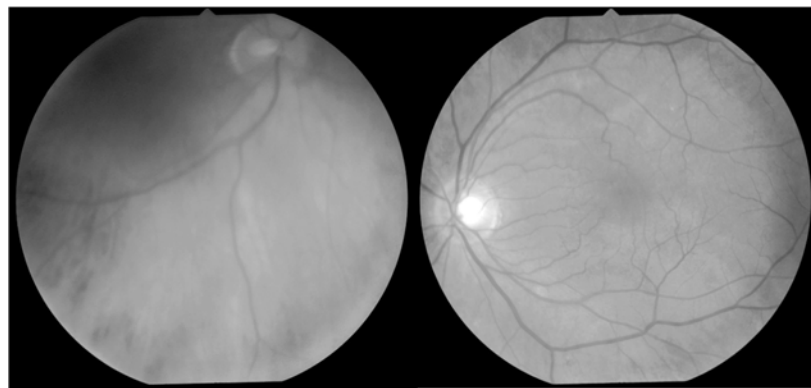


Figure 3 Fundus photograph shows diffuse RPE atrophy in the lower part of right retina. The left fundus is normal. (รูปสีท้ายเล่ม)



Figure 4 Extraocular movement demonstrates a marked limitation of right eye on upward gaze. (รูปสีท้ายเล่ม)

Ocular motility testing revealed hypodeviation of the right eye. He had a marked limitation of right eye movement on upward gaze (Figure 4). The forceduction test was positive for right inferior rectus restriction.

Goldmann perimetry showed a generalized constriction of visual field in the right eye (Figure 5). Conventional electroretinogram demonstrated a moderate reduction of rod and cone responses in the right eye. The patient had normal pattern-reversal visual evoked potentials. There was no evidence of vascular occlusion or leakage during fundus fluorescein angiogram.

Magnetic resonance imaging (MRI) of orbit and brain demonstrated right enophthalmos, right optic nerve tortuosity and crowded right orbital apex (Figure 6A). There was no abnormality of the optic nerve signal and no intracranial lesion. Three-dimensional computerized tomography (3D CT) reconstruction with volume rendering technique (VRT) of facial bones demonstrated hypoplasia of the right facial bones (Figure 6B)

Discussion

Diagnosis of Parry-Romberg syndrome is based on the clinical pictures of progressive hemifacial

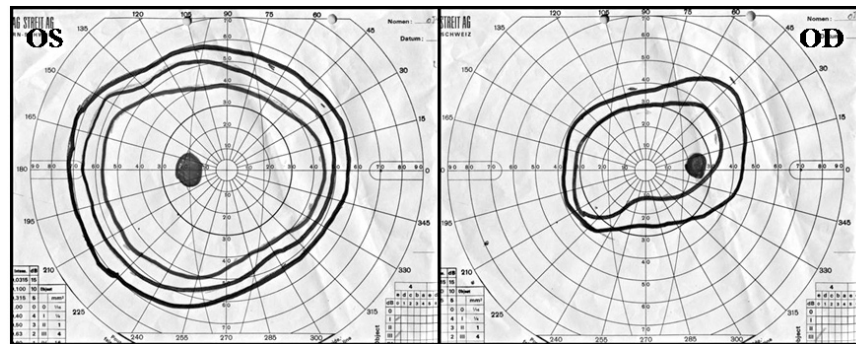


Figure 5 Goldmann visual field testing shows a constricted visual field in the right eye.

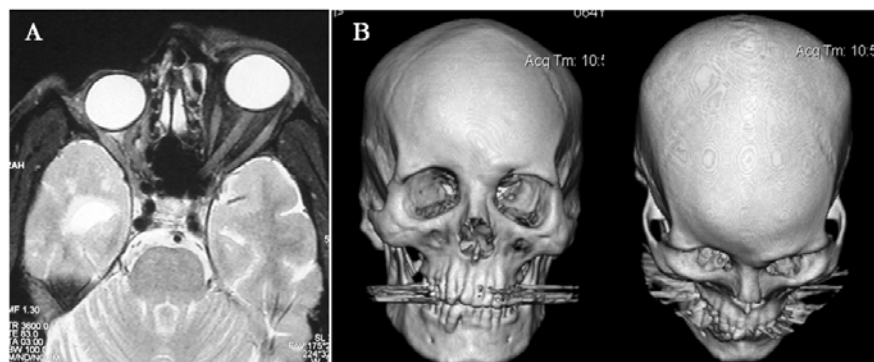


Figure 6 A. MRI of orbit shows right enophthalmos and optic nerve tortuosity.

B. 3D CT reconstruction with VRT shows hypoplasia of right facial bones. (รูปสี่เหลี่ยม)

atrophy. This syndrome is often confused with scleroderma and some authors suggest that the disease might be a form of scleroderma. The criteria for distinguishing these two disorders have been proposed.⁸

Ophthalmologic disorders in Parry-Romberg syndrome are commonly reported in 10-35% of cases^{5,6}. The most common ocular finding are progressive enophthalmos secondary to loss of orbital tissue and atrophic changes of eyelids and eyebrows⁷.

Other ocular abnormalities such as ptosis, iris heterochromia, iridocyclitis, cataract, and glaucoma may be present. Pupillary disorders, causing mydriasis or miotic with poor pupillary light response have been reported including Horner syndrome, Adie's tonic pupil, and Argyll Robertson-like pupil.^{6,7}

Retinal vascular disorders, papillitis and various retinal alterations have also been published in Parry-

Romberg syndrome. Our patient had ipsilateral retinal and RPE disorders which were demonstrated by fundusoscopic finding and electroretinogram. The constriction of visual field would be likely explainable by retinal dysfunction. However, the authors could not find evidence of retinal vasculitis or ischemia in this case.

Ocular motility disturbances including restrictive myopathy, and paralytic strabismus from ocular motor nerve dysfunctions can be found. The presence of restrictive myopathy, as shown in our case suggests that an orbital inflammatory process might be part of this poorly understood syndrome.⁹

Other neurologic disorders such as seizure, cranial nerve disorders, encephalitis, migraine, trigeminal neuralgia, and hemiplegia have been reported in 15% of patients.^{2,7}

There is no specific treatment for Parry-Romberg syndrome. Anti-inflammatory drugs including steroid and other immunosuppressive drugs might have benefit in patients with active inflammatory process. Facial and oculoplastic surgery is indicated in some patients to improve cosmetic disfigurement. However, it is recommended to postpone any operative procedure until no further progression of the disease.⁴

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