A Young Stroke Patient with Bardet-Biedl Syndrome: A casereport

Erh-Chun Chen
Tien-Wen Chen
Ming-Cheng Weng
Chia-Ling Lee

Follow this and additional works at: https://rps.researchcommons.org/journal

Part of the Rehabilitation and Therapy Commons

Recommended Citation
Chen, Erh-Chun; Chen, Tien-Wen; Weng, Ming-Cheng; and Lee, Chia-Ling (2012) "A Young Stroke Patient with Bardet-Biedl Syndrome: A casereport," Rehabilitation Practice and Science: Vol. 40: Iss. 2, Article 7. DOI: 10.6315/2012.40(2)07 Available at: https://rps.researchcommons.org/journal/vol40/iss2/7

This Case Report is brought to you for free and open access by Rehabilitation Practice and Science. It has been accepted for inclusion in Rehabilitation Practice and Science by an authorized editor of Rehabilitation Practice and Science. For more information, please contact twpmrscor@gmail.com.
A Young Stroke Patient with Bardet-Biedl Syndrome: A Case Report

Erh-Chun Chen,1,2  Tien-Wen Chen,2  Ming-Cheng Weng,2  Chia-Ling Lee1,3

1Department of Physical Medicine and Rehabilitation, Kaohsiung Municipal Hsiao-Kang Hospital, Kaohsiung;
2Department of Physical Medicine and Rehabilitation, Kaohsiung Medical University Chung-Ho Memorial Hospital, Kaohsiung;
3Department of Rehabilitation Medicine, Faculty of Medicine, College of Medicine, Kaohsiung Medical University, Kaohsiung.

Bardet-Biedl syndrome (BBS) is a rare inherited disease. Up to date, there have been 14 known BBS-related mutation genes, resulting in ciliopathy with multiple abnormalities such as obesity, pigmentary retinopathy, polydactyly, learning disabilities, poor coordination, renal dysfunction, diabetes and hypertension. This case was a 29-year-old male, who was diagnosed as Bardet-Biedl syndrome since childhood and then was followed up for control of complications. He suffered from stroke at age of 29. Risks of young stroke were surveyed. Poorly controlled hypertension, hyperlipidemia, hyperglycemia and hyperfibrinogenemia were noted. No other examination revealed abnormality. He had received treatment of stroke in acute phase, followed by inpatient rehabilitation programs. Because the retinopathy resulted in visual loss, visual compensation was not possible for him. Besides, BBS-associated poor coordination caused difficulty in rehabilitation. These features challenged to the rehabilitation team. Fortunately, this patient received significant improvement of motor function after rehabilitation training programs. Young stroke could cause significant morbidity and disability. Therefore, we suggest patient with BBS to receive early prevention of stroke and control of complications. We discuss the possible relationship of stroke in young patients and BBS, with particular consideration in rehabilitation management. (Tw J Phys Med Rehabil 2012; 40(2): 103 - 107)

Key Words: Bardet-Biedl syndrome, stroke, rehabilitation

INTRODUCTION

Bardet-Biedl syndrome (BBS) is an inherited ciliopathy due to autosomal recessive transmission, resulting in multiple abnormalities.[1] The primary clinical features are obesity, pigmentary retinopathy, post-axial polydactyly, polycystic kidneys, hypogenitalism, learning disabilities, and renal dysfunction. There are also secondary clinical features including diabetes, hypertension, congenital cardiopathy, Hirschsprung disease, etc.[1,3] The presence of four primary features or three primary fea-
tures plus two secondary features is of diagnostic.\textsuperscript{[2]} Individual clinical phenotype is highly heterogeneous caused by gene mutation of 14 known BBS-related genes.\textsuperscript{[3]}

Among the nonconsanguineous populations of Northern Europe and America, the prevalence ranges from one in 100,000 (North America) to one in 160,000 (Switzerland). Among the Bedouin peoples of Kuwait, where consanguinity is frequent, the prevalence is estimated at one in 13,500.\textsuperscript{[1]}

Young adults are variously defined in published studies as aged less than 40, 45, 50, or 55 years. Ferro et al defined arterial ischemic stroke in young adults as brain infarction at age younger than 55 year-old, the upper age limit.\textsuperscript{[4]} Etiologies are various, including vasculopathy, cardioembolism, metabolic disorders, hypercoagulation status, connective tissue disorders, hematological disorders, smoking, alcohol misuse, drug abuse, migraine, infections, premature atherosclerosis, and hypertension.\textsuperscript{[4]} Other rare causes are monogenic diseases, like Fabry disease, or hereditary endotheliopathy with retinopathy, nephropathy, and stroke (HERNS).\textsuperscript{[5]} Because the presence of risk factors for stroke increases recurrence risk,\textsuperscript{[5]} clinicians should conduct a thorough investigation for possible cardiac, vascular hematologic and genetic risk factors in all patients.

Here we report a male patient with BBS, who suffered from stroke at a young age. We also discuss the possible relationship between BBS and young stroke and consideration in further rehabilitation management.

\textbf{CASE DESCRIPTION}

This 29-year-old male was diagnosed as Bardet-Biedl syndrome by clinical manifestations since childhood. His polydactyly was the initial sign at birth with four rudimentary digits, each one existing at four extremities respectively. Surgical interventions with excision of the accessory digits were performed twice at infant and at age of five. The hand appearance and function were both preserved. Besides, pigmented retinopathy, obesity, diabetes mellitus, hypertension, and mixed dyslipidemia had been diagnosed during the teenage years. He had received regular follow-up at a pediatric outpatient department for over two decades.

He suffered from acute onset of left limbs numbness and weakness on January 8, 2011. In the emergency room, stroke was impressed according to focal neurological signs. Brain-computed tomography showed no intracranial hemorrhage, nor space occupying lesion. Brain magnetic resonance imaging revealed acute lacunar infarction at right paramedian pons (Figure 1). Poorly

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{brain_mri.png}
\caption{Brain magnetic resonance imaging was performed four days after the acute stroke. High signal intensity of right paramedian pons (white arrow) was detected on diffusion-weighted imaging (A) and T2-weighted imaging (B).}
\end{figure}
controlled hypertension, hyperlipidemia, and hyperglycemia were noted then. Survey for stroke risk was performed. Electroencephalogram was indicative of diffuse cortical dysfunction. The nerve conduction study revealed axonal type polyneuropathy. Holter’s scan revealed sinus rhythm with intermittent sinus arrhythmia and sinus tachycardia. Cardiac ultrasound showed left ventricle diastolic dysfunction with mild left ventricle systolic dysfunction. Coagulation status was elevated because of increased plasma fibrinogen (471mg/dl; normal: 200-400 mg/dl). Other hematologic and rheumatologic survey was normal, including complete blood count, ESR, prothrombin time, partial prothrombin time, D-Dimer, C3, C4, ANA, rheumatoid factor, Anti-ds DNA, Anti-Ro, Anti-La, Anti-RNP, Anti-Sm, Anti-SCL-70, Anti-Jo-1 Ab). MR angiogram showed stenosis of bilateral vertebral, basilar and right posterior cerebral arteries. Aspirin 100mg PO QD and Dipyridamole 25mg PO TID were prescribed for secondary prevention of ischemic stroke. Besides, his blindness had been noted since adolescence. Visually evoked potential test proved bilateral optic nerve lesion, prechiasmatic site. Due to left side weakness and daily functioning dependence, he was transferred to the rehabilitation ward.

The initial Brünnstrom stage of left upper limb (proximal/distal) was II/ I, and lower limb was III. He was bedridden and under wheelchair care. There was no significant spasticity at limbs. Barthel index was scored as 40: severe dependence. The complication included left shoulder subluxation.

We arranged physical, occupational and speech therapies for him, which emphasized on motor function control, posture, balance training, daily activity training, verbal and oromotor training respectively. After receiving inpatient rehabilitation for three weeks, he could stand up with minimal assistance and ambulate with a quadricane. At discharge, his left upper limb Brünnstrom stage improved to (proximal/distal) III/II. The Barthel index progressed to 60.

**DISCUSSION**

BBS can be definitely recognized in recent years because a molecular diagnosis is possible. Patients with BBS will need multidisciplinary medical care. The renal abnormalities are the main life-threatening features because they can lead to end-stage renal failure and renal transplantation. Retinal dystrophy would lead to progressive vision loss. Besides, obesity would affect many aspects of health in these patients.

Neurologic impairments reported in patients with BBS include ataxic gait, poor coordination, dysdiadochokinesia, inability to perform tandem gait walking, poor two-point discrimination, and diminished fine motor skills. Although recent research reveals that patients with BBS have statistically significant region- and tissue-specific patterns of brain abnormalities, these neuroanatomical defects on brain imaging had limited explanation for neurologic deficits observed in BBS.

Reviewing research which directly links BBS to stroke (searching on Medline and Pubmed), we only found one case report regarding a young stroke patient with BBS. Up to date, no evidence recognizes BBS as the risk factor of young stroke. By reporting this case, we may provide information that young stroke is related to BBS in direct or indirect pathophysiology.

Hypertension, diabetes mellitus, dyslipidemia, obesity and hyperfibrinogenemia are recognized as risk factors of ischemic stroke. Before stroke attack, this patient was under poor control of blood sugar (HbA1C: 13.1), high blood pressure (162/113mmHg), abnormal blood lipid profile (Cholesterol: 231 mg/dL, Triglyceride: 549 mg/dL) and overweight (body mass index: 36.75). Many features of BBS would secondarily cause young stroke in this patient.

The features of BBS may also cause difficulty in rehabilitation in patients with stroke. In planning a rehabilitation program for stroke with BBS, neurologic impairment must be considered, including ataxic gait and poor coordination. Visual impairment was another challenge for the rehabilitation team in this patient since visual compensation was not possible for him. Fortunately, this patient received significant improvement of motor function after rehabilitation training programs.

**CONCLUSION**

Patients with BBS seem to have greater risk of young stroke although the primary and secondary etiologies are not clarified and degree of relationship is not
fully established. Young stroke could cause significant morbidity and disability. Therefore, the issue of stroke prevention in patients with BBS may be raised for physicians. Physiatrists should focus more on posture, balance, and coordination training in patients with complex disabilities associated with Bardet-Biedl syndrome.

REFERENCES

巴德-畢德氏症候群併發年輕型腦中風：病例報告

陳爾駿 1,2  陳天文 2  翁銘正 2  李佳玲 1,3

高雄市立小港醫院復健科 1 高雄醫學大學附設中和紀念醫院復健科 2
高雄醫學大學醫學院醫學系復健醫學科 3

巴德-畢德氏症候群為一少見的遺傳性疾病，現已經發現缺陷的十四處基因，主要造成纖毛病變，臨床上病人有多重功能異常，例如肥胖、色素性視網膜病變、多指趾畸形、學習障礙、動作協調性異常、腎功能不佳、糖尿病和高血壓。

此病例為一名二十九歲的男性，出生後即診斷為巴德-畢德氏症候群，隨後在小兒科追蹤並控制併發症。在二十九歲時發生年輕型梗塞性腦中風，而中風的危險因子檢查，除了血糖、血壓及血脂控制不佳，及纖維蛋白原上升，其他檢查並無異常，經急性期的中風治療，繼續接受住院復健。此病患因為視網膜病變造成雙眼失明，視覺無法代償動作的控制，並且巴德-畢德氏症候群合併的動作協調性異常，不利於運動訓練，這都是對復健治療團隊的挑戰。經復健後病人的運動功能及日常生活功能均有明顯進步。

中風造成病人明顯的障礙，故建議對於巴德-畢德氏症候群必須及早預防中風及控制併發症。在此我們也討論年輕型中風與巴德-畢德氏症候群的可能相關性，以及復健處置的特殊考量。（台灣復健醫誌 2012；40(2): 103 - 107）

關鍵詞：巴德-畢德氏症候群(Bardet-Biedl syndrome)，中風(stroke)，復健(rehabilitation)