

significant morbidity with selective cannulation of the intercostal arteries, lumbar arteries and the vertebral arteries.

Interventional neuroradiology has an important role in the treatment of SDAF. The first case was treated with endovascular embolisation which halted the progression

of the paraparesis. Unfortunately, the second patient declined treatment.

In conclusion, spinal dural arteriovenous fistula is a potentially treatable cause of myelopathy. Spinal angiography remains the gold standard for confirming or refuting the presence of SDAF.

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# Psychiatric Presentation of Huntington's Disease in a Malaysian Family

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## Summary

A 32-year-old Chinese lady presented to the Psychiatric Clinic with a history of change in personality for 2 years and abnormal movements for a year. After thorough investigations and observation a diagnosis of Huntington's Disease was made. Her elder brother was traced and found to have Huntington's Disease as well. He had a long standing history of antisocial behaviour and substance abuse long before the onset of the choreiform movements. Her younger brother also has choreiform movements for the last 2 years and had recent change in personality. Their mother also had abnormal movements and was recorded to be depressed and attempted suicide. The maternal grandfather had a mental illness and was warded at a mental institution till his death in 1942. Psychiatric presentation of Huntington's Disease in this Malaysian family is prominent and preceded the characteristic movements in the present generation.

**Key Words:** Huntington's Disease, Psychiatric symptomatology

### Introduction

Huntington's Disease (HD) is an autosomal dominant disorder. The site of genetic abnormalities has been identified on the short arm of chromosome 4. It is a serious disorder that manifests in adulthood, when the affected individuals may have passed down the defective genes to half of their offspring. Huntington's Disease presents with movement disorder and dementia. However, the first descriptions by George Huntington in 1872 describe a triad of suicide, hereditary tendency and onset in adult life. Descriptions of HD mainly concentrated on the neurological aspects but equally important are the psychiatric complications. Psychiatric disorder is common in HD and are known to precede the neurological changes. This is a report of a Malaysian family with HD in which the psychiatric symptomatology presented prominently.

### Case Report

A 32-year-old Chinese lady presented at the Psychiatric Clinic in December 1992. Her uncle noticed a change in her personality for about 2 years. She became withdrawn, argumentative, restless and had poor sleep at night. She left her job as a coffee shop waitress and stayed at home with her uncle. She was noted to be irritable and would go out of the house at odd hours of the day and night. On occasions she was found wandering in town. A year later she developed jerking movements of her hands and had abnormal gesturing on and off. On the day of presentation she became irritable and broke the windows in the house.

She was admitted for observation. Relevant features were choreiform movements of the limbs and slurred speech. Her recent memory was poor and she was not orientated to time and place. There were no delusions or hallucinations. Investigations included a full blood picture which was normal, serum copper of 23.5 (10.2-26.0), serum caeruloplasmin 0.43 (0.15-0.60), negative HIV screening LE and RH factor. Liver function and renal profile were within reference range. A CT scan of the brain was reported "prominent subarachnoid spaces suggestive of generalised cerebral atrophy with loss of curvature of the head of the caudate nucleus suggesting atrophy". A mini mental state produced a score of 8/30 which is a poor score and suggested cognitive impairment.

The EEG was reported "background activity is very suppressed giving rise to an abnormal EEG which is flat and featureless in all leads".

In the family history, her maternal grandfather had a mental illness and was admitted to the mental hospital at Tanjung Rambutan (now known as Hospital Bahagia). He died there in 1942. Unfortunately we could not obtain any record of his illness nor if he had any abnormal movements. The family originated from Fujian Province in China and speak Hokkien dialect. There is no history of intermarriage with other races.

Her mother had a history of involuntary movements of all limbs, progressive loss of memory and abnormal behaviour starting from the age of 23 years. She presented at University Hospital 5 years later and was noted to have poor recent memory, unsteady gait and choreiform movements of all 4 limbs. She was diagnosed as having Huntington's Chorea. Her symptoms progressively worsened. After her husband died of a stroke in 1972, she was reported to be depressed and attempted suicide twice by jumping into a well. She stopped eating and died sometime in 1975.

Her older brother is 34 years old. He has problems since young and was said to be playful and lacked concentration. His parents could not control him and he was taken care of by his paternal grandmother. He stopped schooling at Form 1 and has been involved in petty crime. He started abusing drugs at the age of 16. He took cannabis and heroin and had been using the intravenous route at times. He has been jailed twice and sent for rehabilitation at Pusat Serenti 3 times. He was traced and examined. He had an ataxic gait with choreiform movements of his limbs. His speech was slurred but his recent and remote memory was intact. Subsequently he was admitted to the Psychiatric Ward, having been brought in by the Police for throwing stones in public. He was on heroin at the time and there were no psychotic features noted. He was investigated and of relevance was that he tested HIV positive.

She has a younger brother who was recently released from detention in Taiwan for working illegally there. He has slurred speech and choreiform movements

when seen recently. The uncle also reports a change in his personality in that he is more impulsive and likes to go out wandering.

**Progress :** The 32-year-old lady progressively deteriorated. Her movements became worse and she had frequent falls. Her memory also deteriorated and she developed bed sores at a private nursing home. She died of septicemia in April 1994. A limited post mortem revealed focal cerebral atrophy involving frontal and occipital regions. Histology showed increased gliosis in the region of the caudate and putamen nuclei.

The 34-year-old man was sent to Hospital Bahagia Ulu Kinta. He refused to go for drug rehabilitation and kept insisting on discharge. Blood samples from both brothers have been sent for confirmation of HD by molecular genetics.

## Discussion

In the 32-year-old lady, her psychiatric symptoms preceded the neurological manifestations by about a year. There were gross changes in her personality which was noticed by her uncle. This led to her loss of job and her irritability and impulsive behaviour led to difficulties with her uncle with whom she stayed with. This is correlated by the post mortem which showed atrophy at the frontal lobe. Frontal lobe involvement is known to produce the above disturbances<sup>1</sup>.

The 34-year-old man has antisocial personality in addition to his heroin dependence. Persons who subsequently developed Huntington's Disease have been well known to have marked difficulties even before their onset of neurological signs<sup>2</sup>. However he has not yet shown signs of dementia. The younger brother has no premorbid antisocial traits but has prominent movement disorder. Like his sister he has shown a change in personality with marked impulsivity and irritability at times.

In contrast the 32-year-old lady and her mother showed signs of dementia and rapidly deteriorated. The mother died 5 years after being first diagnosed at the age of 30. The 32-year-old lady died at an earlier age and her

dementia was rapid. When first seen, she had mild memory deficits but on follow-up she rapidly deteriorated and died within 16 months. This finding is not in keeping with the theory that maternal transmission confers a protective effect and the HD is milder and shows up much later in life than paternal transmission<sup>2</sup>.

The pathological changes involve the caudate nucleus. This is supported by the CT scan which suggested atrophy and by post mortem histology which showed increased gliosis and cellularity in the region of the caudate and putamen nuclei. The findings are in keeping with a diagnosis of HD<sup>1</sup>.

In view of the autosomal dominant inheritance, it is essential to trace and examine all family members. The uncle who has been the informant is asymptomatic. He has been counselled. Efforts are made to see the younger brother. The older brother is insightful and is aware that he is also HIV positive as well as having an incurable deteriorating disease. He seems determined to resume his heroin dependence till he dies.

The grandfather has mental illness though the exact nature is not clear. It must be of psychotic proportions for him to be admitted to the mental hospital in the 1930s. Unfortunately, we do not have any documentation on his illness as the suspicion is that he carried the gene.

In this family, the psychiatric aspects of Huntington's Disease is prominent and at times preceded the neurological signs. Although the first Malaysian report of HD has been recent<sup>3</sup>, with more awareness, more cases will be picked up and help to the families involved be rendered to reduce transmission.

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# Bladder Outflow Obstruction Masquerading as Pelviureteric Junction (PUJ) Obstruction

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## Summary

We report a case of bladder outflow obstruction presenting with upper tract dilatation mistaken initially as pelviureteric junction (PUJ) obstruction. The lower tract obstruction ought to be dealt with first before upper tract obstruction is assessed because the renal pelvic pressure is significantly affected by vesical filling and high bladder pressure.

*Key Words:* Bladder outflow obstruction, Pelviureteric junction obstruction

## Introduction

Patients with bladder outflow obstruction commonly present with obstructive symptoms such as hesitancy and poor stream. In this case the patient developed upper tract symptoms associated with renal impairment. Due to the tendency to develop upper tract dilatation, the dilated renal pelvis might be mistakenly construed as pelviureteric junction obstruction.

## Case Report

Mr. C.S. is a 59-year-old gentleman who was referred to the Department of Urology Hospital Kuala Lumpur for the management of a solitary right kidney with hydronephrosis and renal impairment. He gave a 9-

year history of intermittent left loin ache for which he usually consulted his general practitioner. In April 1994, he sought treatment in a private hospital in Ipoh for severe acute left renal colic associated with fever and vomiting. Further investigations with intravenous urography and retrograde pyelography demonstrated his left kidney to be non-functioning & grossly hydronephrotic associated with a narrow pelviureteric junction (Fig. 1); the right pelvicalyceal system appeared normal. At exploration there was pyonephrosis and a simple nephrectomy was undertaken based on the assumption of a left non-functioning and pyonephrotic kidney with pelviureteric junction (PUJ) obstruction. His postoperative recovery was uneventful.