Case Report

Missed diagnosis: A case of Von Hippel-Lindau that highlights importance of considering cancer syndromes in multiple system symptoms

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ABSTRACT

Von Hippel-Lindau disease is an autosomal dominant trait that predisposes individuals to benign and malignant tumors. It is rare, with prevalence of around 1 in 36,000. This case reports systematic limitations in knowledge of cancer syndromes, and provides a resource for junior doctors, as well as offering an interesting look at complex diagnostics.

Key words: Cancer, syndromes, Von Hippel-Lindau

INTRODUCTION

Von Hippel-Lindau (VHL) disease is an autosomal dominant trait that predisposes individuals to benign and malignant tumors. It is rare, with prevalence of around 1 in 36,000.[1] The most common tumors in VHL are central nervous system (CNS) and retinal hemangioblastomas, clear cell renal carcinomas, pheochromocytomas, pancreatic neuroendocrine tumors, pancreatic, and cysts. VHL results from a mutation in the VHL tumor suppressor gene on chromosome 3p25.3.[1,2] We use this case to highlight the rarer cancer syndromes and to provide a document offering some brief review information.

CASE REPORT

A 38-year-old unemployed gentleman was admitted through accident and emergency due to a fall after prolonging and worsening muscle tone. There had been increasing incoordination and difficulty walking for a year; he has recently been seen by the neurologists and nephrologists following general practice (GP) referral. He now has a known cancer syndrome, but at the time of accident and emergency presentation this had not been diagnosed.

He first noticed his loss of muscle tone a year ago and went to his GP who referred him to the nephrologists, as he was also complaining of a constant dull ache in his abdomen and to the neurologists. The neurologists sent him for a scan. At this time, he was a builder and worked at high levels in London on scaffold. He began to find himself wobble a bit and was signed off work. As he was self-employed, he now has no further employment.

He has been seen by a nephrology junior doctor regarding his neurology requested scan which showed a CNS lump and a kidney lump [Figures 1 and 2].[4,5] The junior proceeded to tell the patient it appeared his cancer had spread. This patient at the point of contact with us believes he has a primary in his kidney, which has spread to his brain. The patient has been a smoker of 10 a day for 22 years, is an ex drinker - stopped when became dizzy. His father had had an operation on his kidney, but had died in a road traffic accident 6 months after. This was 30 years ago. No other history of note. He currently takes diclofenac.

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The patient was well with tenderness over the left thigh. An X-ray was carried out and no fracture was identified. However, the patient was admitted to neurosurgery with a urology and nephrology request for input.

DISCUSSION

This case is interesting as it shows the importance of broad knowledge in the clinician. It shows the relevance of cancer syndrome knowledge and reinforces multisystem diagnostics.

The dizziness is suggestive of cerebellar involvement, and the history is suggestive of a progressive disease. This relates to a CNS hemangiobalstoma, of which 80% occur in the cerebellum and frequently forms part of the VHL phenotype.

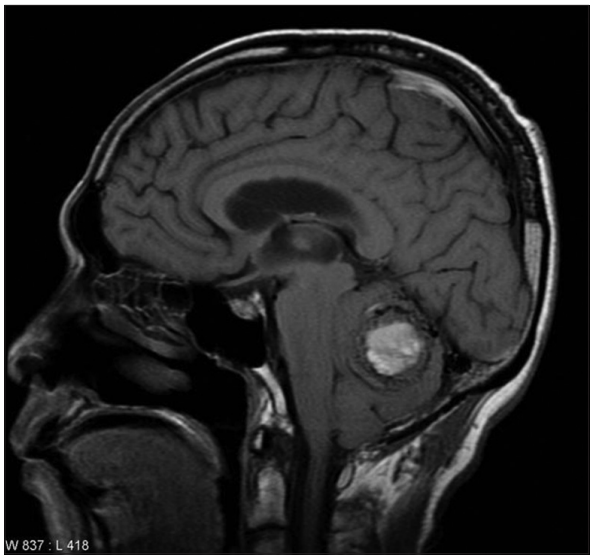

Figure 1: Magnetic resonance brain T1 - central nervous system haemangioblastoma visible

Whilst a kidney biopsy has not yet been undertaken, a clear cell cancer on biopsy would further point to a VHL diagnosis, and it's important to note the father had died prematurely, but already had a history of previous kidney operation.

This patient had at one point been told his kidney cancer had metastasized and that what was causing his signs. This has a far bleaker outlook than what was in fact two primaries.

This had a massive psychiatric effect on the patient, thus an important professionalism point point is before breaking any news particularly bad, make sure you are correct.

Other cancer syndromes frequent presentations are briefly shown in Table 1.

CONCLUSION

Cancer syndromes have a substantial impact on patient's lives, careful management and consideration must be given to these. They may occur as multisystem disorders, and as

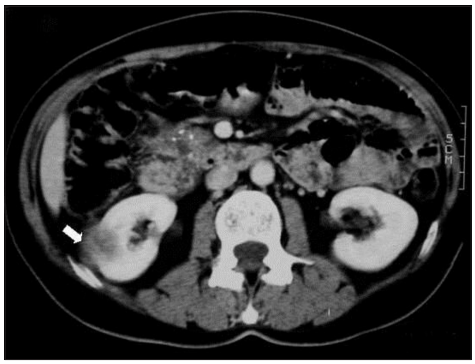

Figure 2: Computed tomography of abdomen demonstrating a kidney mass

Table 1: Some cancer syndromes and their associated solid lesions[1-3]

Syndrome	Common tumours	Other tumours
Fanconi anaemia (recessive)	Manifestations related to haematopoiesis Aplastic anaemia Myelodysplastic syndrome Acute myeloid leukaemia	Hepatic tumours and squamous cell carcinomas of the oesophagus, oropharynx and uvula are solid tumours commonly linked to Fanconia Anaemia
Familial adenomatous polyposis (dominant)	Benign adenomas throughout colon	Other tumours increased in frequency include; osteomas, adrenal adenomas and carcinomas, thyroid tumours and desmoid tumours
Hereditary nonpolyposis colon cancer (dominant)	Increases the risk of colorectal cancer	Other cancers are increased in frequency. These include; endometrial cancer, stomach cancer, ovarian cancer, cancers of the small bowel and pancreatic cancer
Li-Fraumeni syndrome (dominant)	Multiple independent primary cancers	Cancers linked to this disorder include; soft tissue sarcomas (often found in childhood), osteosarcoma, breast cancer, brain cancer, leukaemia and adrenocortical carcinoma
Xeroderma pigmentosum (recessive)	The risk of skin cancer is more than 10,000 times that of normal individuals and includes many types of skin cancer, including melanoma and nonmelanoma skin cancers	

such, these syndromes should always be considered in such examples.

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