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. 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. 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]. He 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 hemangioblastoma, of which 80% occur in the cerebellum and frequently forms part of the VHL phenotype.

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

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Common tumours</th>
<th>Other tumours</th>
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<tbody>
<tr>
<td>Fanconi anaemia (recessive)</td>
<td>Manifestations related to haematopoiesis</td>
<td>Hepatic tumours and squamous cell carcinomas of the oesophagus, oropharynx and uvula are solid tumours commonly linked to Fanconi Anaemia</td>
</tr>
<tr>
<td>Familial adenomatous polyposis</td>
<td>Aplastic anaemia, Myelodysplastic syndrome</td>
<td>Other tumours increased in frequency include; osteomas, adrenal adenomas and carcinomas, thyroid tumours and desmoid tumours. Other cancers are increased in frequency. These include; endometriat cancer, stomach cancer, ovarian cancer, cancers of the small bowel and pancreatic cancer. Cancers linked to this disorder include; soft tissue sarcomas (often found in childhood), osteosarcoma, breast cancer, brain cancer, leukaemia and adrenocortical carcinoma</td>
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<tr>
<td>(dominant)</td>
<td>Acute myeloid leukaemia</td>
<td></td>
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<td>Hereditary nonpolyposis colon</td>
<td>Benign adenomas throughout colon</td>
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<tr>
<td>cancer (dominant)</td>
<td>Increases the risk of colorectal cancer</td>
<td></td>
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<td>Li-Fraumeni syndrome (dominant)</td>
<td>Multiple independent primary cancers</td>
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<td>Xeroderma pigmentosum (recessive)</td>
<td>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</td>
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such, these syndromes should always be considered in such examples.

REFERENCES


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