**Beckwith-Wiedemann Syndrome and Bilateral Phaeochromocytoma: a Diagnostic Challenge**

**Background**

Beckwith-Wiedemann Syndrome (BWS) is a rare overgrowth disorder secondary to mutations in growth-regulatory genes on chromosome 11p15.5. A well-established association exists between BWS and benign and malignant tumours, most commonly Wilms’ tumour and hepatoblastoma. We describe a patient with BWS and bilateral phaeochromocytoma/paraganglioma (PPGL), which is much less well described.

**Initial presentation**

- A 14-year-old girl with genetically confirmed BWS presented with headaches, sweating, palpitations and hypertension (BP 177/117)
- Previously routine screening had detected a bladder rhabdomyoma (aged 2 years) and a pancreatoblastoma (aged 7 years), both surgically resected.
- Biochemistry was in keeping with a diagnosis of phaeochromocytoma (Table 1) and an abdominal MRI demonstrated bilateral complex adrenal cysts ≤ 2cm (Figure 1), however iodine-123-meta-iodobenzylguanidine (MIBG)-scan showed no abnormal sites of activity.

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>24 hr urine catecholamines</td>
<td>Normetadrenaline 1.6 umol</td>
<td>0.6-3.5</td>
</tr>
<tr>
<td></td>
<td>Metadrenaline 0.1 umol</td>
<td>0.2-1.4</td>
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<tr>
<td>24 hr urine catecholamines</td>
<td>Normetadrenaline 22.3 umol</td>
<td>0.6-3.5</td>
</tr>
<tr>
<td></td>
<td>Metadrenaline 0.8 umol</td>
<td>0.2-1.4</td>
</tr>
<tr>
<td>Plasma catecholamines</td>
<td>Normetadrenaline 9514 pmol/L</td>
<td>120-1180</td>
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<tr>
<td></td>
<td>Metadrenaline 295 pmol/L</td>
<td>80-510</td>
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</tbody>
</table>

**Management**

- A personalised management plan was created using the BSPEd endorsed Paediatric Endocrine Tumours consensus and the adult Phaeochromocytoma and Paraganglioma guideline
- She was commenced on the alpha-blocker Doxazosin 1mg once daily as an outpatient, then admitted for BP optimisation using phenoxybenzamine. This dose was gradually increased under close monitoring until complete alpha-blockade was achieved.
- Large volumes of intravenous fluids were necessary pre-operatively for volume expansion.
- Following successful bilateral adrenalectomies, histological analysis confirmed phaeochromocytoma.

**Post-operative course**

- Hydrocortisone was initially commenced intravenously intra-operatively, then converted to oral hydrocortisone (10 mg morning, 5 mg at lunchtime, 5 mg evening) and fludrocortisone 100mcg once daily once tolerated.
- Despite successful bilateral adrenalectomies she remained hypertensive (systolic BP >130) and further investigation found plasma normetanephrine remained elevated (4152pmol/L).
- A Gallium-68 DOTANOC PET-CT scan showed increased uptake in para-caval and left para-aortic lymph nodes and in the pancreas (Figure 3).

**Learning points:**

- This case demonstrates the importance of on-going vigilance for tumour development in patients with BWS.
- Phaeochromocytomas not detected on MIBG-scanning are more likely to be metastatic and due to SDHB mutations, which carry an unfavorable prognosis. This case highlights the importance of undertaking further imaging after a negative MIBG scan if the clinical and biochemical picture is highly suggestive of phaeochromocytoma.
- An updated guideline on the management of paediatric PPGLs is needed.

**References**

2. Paediatric Endocrine Tumours. Editor Spoussens H. BSPEd and UKCSG, published October 2005

There are no conflicts of interest to declare.