Diagnosis and management of BHD-associated kidney cancer (Stamatakis *et al*., 2013)

Lay Summary

Who wrote this paper?

This paper was written by Dr W. Marston Linehan and his team at the National Cancer Institute, at the National Institutes of Health in the United States. Dr Linehan and his team have nearly twenty years of experience in treating BHD kidney cancer.

What is this paper about?

This paper recommends steps to diagnose and manage kidney cancer caused by BHD syndrome.

What does the paper say?

Once a patient has been diagnosed with BHD, baseline chest and abdominal scans should be performed to see how badly affected the patient's lungs and kidneys are. Chest CT should be used to identify lung cysts or small pneumothoraces. Regular lung scans are not necessary, unless the patient has symptoms requiring medical treatment.

Abdominal CT or MRI (with contrast) should be used to assess the kidneys; ultrasound scanning often misses BHD renal tumours as the tumours reflect sound waves similarly to surrounding healthy tissue. Patients without any renal tumours at this initial scan should have their next scan in 36 months. Patients with renal tumours should have scans more regularly. How often will depend on how many tumours they have and how fast the tumours are growing.

Roughly 3 in 10 (27%) of BHD patients are found to have kidney tumours at their initial scan, and of those, 65% (2 in 3) have more than one tumour.

Once the largest tumour is 3 cm in diameter, all tumours in that kidney should be surgically removed. If both kidneys have tumours larger than 3 cm, tumours will be removed in two separate surgeries.

Before surgery, the patient should have a pulmonary assessment and excessive positive pressure ventilation during surgery should be avoided to reduce the risks of the patient developing a pneumothorax during the operation.

Where possible, partial nephrectomy is preferable to total nephrectomy, as patients may develop more tumours over time. If necessary, the patient should be referred to a center of excellence with experience of performing partial nephrectomies. Lymph node dissection is not normally performed unless pre-operative scans suggest that the disease may have spread.

Ablative techniques, such as cryoablation or radiofrequency ablation are not recommended for BHD patients, as they can make any future surgeries more difficult. However, these techniques may be appropriate for older patients, or for those who cannot undergo surgery.

On average, BHD patients who had surgery at NCI had 5 tumours removed during surgery. Tumours were most commonly of mixed chromophobe and oncocytic histology. However, chromophobe, oncocytoma, clear cell and papillary cell tumours have also been found. Clear cell tumours tend to be more aggressive than other tumour types.

When managed in this way, BHD patients have an excellent prognosis, and most patients will only need one surgery in their lifetime to manage their kidney disease. In the few cases where BHD kidney tumours have spread, tumours were of clear cell histology, or the disease was very advanced by the time it was diagnosed.

The youngest BHD patient who had kidney cancer was 24, so diagnostic genetic testing at 21 years old is recommended.