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ORIGINAL ARTICLE

Birt–Hogg–Dubé: beyond the clinical manifestations

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Abstract Clinicians and scientists understand the medical implications of BHD; however, what may not be apparent to clinicians and scientists are the psycho-social aspects of living with BHD. Although medical reality differs among people who have Birt–Hogg–Dubé, they often share multiple non-medical ramifications ranging from economic and physical insecurity to interruptions in familial communication patterns and relationships. Physicians cognizant of the psycho-social aspects of having BHD are in a position to offer enhanced and meaningful non-medical interventions and care to their patient with BHD.

Keywords Renal cancer · Birt–Hogg–Dubé · Pneumothorax · Fibrofolliculoma · Psycho-social

Individuals and their families, who live with Birt–Hogg–Dubé (BHD), experience a varied medical experience. For some patients, life may be relatively simple involving mild manifestations with periodic kidney imaging, while for others, life may be more difficult: living with multiple unwanted facial skin bumps, repeated trips to the emergency room to endure chest tube placement or undergoing major renal or pulmonary surgery. Clinicians and scientists understand BHD medical implications; however, what may not be apparent to clinicians and scientists are the psycho-social aspects of living with BHD. Although medical reality differs among people who have Birt–Hogg–Dubé, they often share multiple non-medical ramifications ranging from economic and physical insecurity to interruptions in familial communication patterns and relationships. Physicians cognizant of the psycho-social aspects of having BHD are in a position to offer enhanced and meaningful non- medical interventions and care to their patient with BHD.

This paper is not reflective of scientific inquiry, but is derived from a clinician's perspective from 15 years' experience of evaluating, counseling and listening to $\sim$ 350 people who have Birt–Hogg–Dubé syndrome. While some of the issues presented are globally experienced by people who have significant health problems, several are unique to individuals with BHD. Recurrent issues/themes have arisen that are of mild to significant concern to many, resulting in an intensity range of frustration, fear and anxiety. While some patients fare well with few medical or psycho-social ramifications of having BHD, others feel a sense of medical, economic and familial insecurity.

The most frequently voiced concern of patients is that their local physician has never heard of BHD, and the doctor does not know what to do or how to care for them. This fact produces insecurity and fear that malignant kidney tumors will be missed; some people believe or are fearful that their future with BHD includes dialysis and/or premature death. By providing copies of several clinical papers to patients they may share with their local doctors can substantially ease concern.

Patients with significant pulmonary manifestations offer the strongest voice. They question whether their lungs can handle another pneumothorax or whether both lungs will collapse at the same time. Some people fear their life span is threatened by repeated pneumothoraces, and have concern they will develop chronic lung disease, significantly interfering with their quality of life or impact their ability to work. People with significant pulmonary manifestations

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seek a pulmonary team confident in management of BHD lungs. However, there are very few pulmonologists/thoracic surgeons in the United States who have clinical experience managing BHD lungs. Lymphangioleiomyomatosis (LAM), a rare disorder unrelated to BHD, is associated with multiple pulmonary cysts. The LAM Foundation, which is the primary patient advocacy group, supports 26 clinics in hospitals and medical centers throughout the United States. Pulmonologists and thoracic surgeons at these LAM clinics may be a beneficial clinical resource for patients who have significant pulmonary manifestations. LAM clinic locations can be found on the LAM Foundation web-site http://www.thelamfoundation.org/.

While most people who have fibrofolliculomas find their facial lesions a burden, the presence of fibrofolliculomas does not appear to cause social isolation or significant depression. However, patients consistently seek information about what can be done to eradicate their fibrofolliculomas. There is an oft-stated ardent plea to scientists- please find something to make our fibrofolliculomas disappear.

All patients strive to learn the gold standard for kidney and lung surveillance. Establishing a global standard for renal surveillance may be a goal for a future international BHD Symposium. MRI imaging of the abdomen with/without contrast every 2–3 years for gene-positive or at-risk adults is the current recommendation of the Urologic Oncology Branch at the National Institutes of Health. CT scans with contrast is an acceptable form of imaging, however, use of MRI imaging will reduce the lifetime risk associated with radiation exposure. Use of ultrasound for kidney surveillance is a not a reliable method for detecting or following renal masses associated with Birt–Hogg–Dubé. When kidney tumors are present, the frequency and imaging modality is determined by the patient's urologic surgeon.

A frequent question of patients pertains to the age children should undergo genetic testing and initiate kidney surveillance. As kidney tumors have not been identified in minors, the NIH program recommends genetic testing around 18–20 years of age with initiation of renal surveillance in gene positive individuals. Optimum surveillance strategies for pulmonary cysts have not yet been determined.

Perceived and real economic burdens exist for some people living with BHD. There is concern whether health insurance will cover the cost of screening, or whether out-of-pocket expenses associated with kidney tumor surgery, emergency room visits for recurrent pneumothorax, or costs associated with pleurodesis can be personally met. For patients who do not have insurance, surveillance may not be an option and dying from kidney cancer is a lived fear.

Some patients are concerned they will not be able to advance their career or leave their current job as they may lose health insurance benefits with a future employer. For those who are unemployed or job insecure, fear exists that they now have a pre-existing condition, which may jeopardize future insurability. Although health care providers are not experts in state/federal laws, cursory knowledge of and referring patients to a few protections may offer patients some relief and provide guidance for personal exploration. In the U.S. forty-five states have enacted state laws protecting their citizens from genetic discrimination by health insurers; but the state laws vary in their level of protection. Brief summaries of these state statutes can be found though the National Human Genome Research Institute's web-page www.genome.gov/policy. Moreover, one of the primary provisions of the H.I.P.P.A. Act in the United States provides portability of health insurance plans, allowing people to carry their current health policy from one job to another, potentially avoiding a pre-existing clause. Helpful information is available on multiple websites http://www.dol.gov/ebsa/faqs/faq_hipaa_ND.html.

The United States G.I.N.A. Act of 2008 contains language preventing health insurers from engaging in genetic discrimination. Genetic discrimination occurs if people are treated unfairly because of differences in their DNA that increase their chances of getting a certain disease. GINA also includes language precluding employers from using a person's genetic information in decisions about hiring, firing, job assignments or promotions http://www.genome.gov/10002328. Referral to some of the above cited sources or providing a copy of this paper may provide some comfort to patients.

An area of considerable concern to many is the implications to their family and a potential for family disruption. Familial communication patterns differ among families. Some families know a great deal about one another; there is frequent sharing of daily life events between parents, children, brothers, sisters, aunts, uncles, cousins. Alternatively, it is the cultural norm in some families to communicate rarely about personal daily life events, especially about health issues. For patients in these families, communicating about an inherited, potentially lethal disorder is a daunting and difficult responsibility.

Most parents express mild to extreme discomfort and difficulty in communicating genetic risk to their offspring; rarely patients do not share any information with family members. It appears that uptake of genetic testing and initiation of surveillance of at-risk family members occurs more frequently in families in which the affected members have had significant morbidity. However, this phenomenon has not been systematically studied in the BHD population.

Concern is expressed among some patients that their family members are not sufficiently motivated to pursue genetic testing and kidney surveillance. Many patients share medical papers, handouts, and inform family about

the process to obtain genetic testing, but family members do not take action. In these situations worry and fear about the health and well being of their family members is amplified.

People with rare disorders often verbalize a sense of isolation; their doctor has never heard of the condition and they do not know anyone who is similarly affected. These two facts result in a feeling of "aloneness". People can often learn more about BHD from one another than from medical professionals. BHD families should be encouraged to attend any and all scientific meetings open to them, as families' welcome sharing their experiences and learning they are not alone. Helpful information is available on the Myrovlytis (MT) website bhdsyndrome.org The website contains patient—specific informational materials and scientific papers are available, as well as a secured internet resource allowing anonymous communication between people and families with BHD.

Cancer Genetic Counselors (CGC) and/or Advanced Practice Nurses in Genetics (APNG) may be useful in helping patients and their families cope with the psychosocial ramifications of BHD. Counselors may offer help and provide solutions with some issues or offer support and guidance in others. Most academic medical centers in the United States have Cancer Genetic Centers with certified genetic counselors or credentialed Advanced Practice genetic nurses. Patients can find professional genetic counseling providers through the National Society of Genetic Counselors (NSGC) www.nsgc.org or the International Society of Nurses in Genetics (ISONG) http://www.isong.org/.

Although a clinicians' primary responsibility is the diagnosis and management of the physical manifestations of BHD, expanding awareness of the broader implications of having BHD provides an opportunity to offer patients an enriched approach to care. A physician's careful listening combined with a cursory knowledge of, and referral to, several resources cited above offers an opportunity to provide an enhanced level of care beyond the clinical manifestations of Birt–Hogg–Dubé.

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