Living with VHL from the Young Adult Perspective

by Lindsay Schmidt, MS, CGG

Von Hippel-Lindau syndrome affects patients in various ways. While most research has focused on the genetics of VHL and medical implications of the syndrome, social and emotional experiences of VHL patients have received far less attention. Additionally, there is a lack of literature addressing the experiences of young adults living with VHL. This lack of knowledge served as inspiration for my thesis study, which I completed at Brandeis University while earning my master’s degree in Genetic Counseling.

The purpose of this study was to explore the social and emotional aspects of living with VHL as a young adult. My investigation was performed by conducting telephone interviews with ten young adults (18–26 years old) living with VHL in the US. After interviewing all participants, I identified five overarching themes to which many VHL patients will relate.

Living with Uncertainty

Many participants expressed that living with uncertainty is a challenging aspect of living with VHL. As Sophie pointed out, a VHL diagnosis leads to many ‘what-if questions’, and participants seemed to struggle with if, when, and how VHL will affect them. Tumor growth, timing of VHL manifestations, and VHL’s impact were concerns.

“Every time I go to NIH, I just never know what I’m going to find out; is there anything new or is anything getting worse?” —Peter

Maintaining a Positive Attitude

Despite living with continual uncertainty, many participants seemed to maintain a positive attitude. Several participants positively framed their VHL experiences while others exhibited perseverance.

“Experiences that you go through either make you better or they make you bitter. I wanted them to make me better, and I think that they have. … it has … been a really awesome growing experience for me that I’m lucky enough to have.” —Julie

Significant Means of Support

Participants reported large support systems, which often included a combination of family, romantic partners, friends, coworkers, and health care professionals. However, when it came to VHL-related concerns, single participants most often reported their mothers as their strongest confidants, while married participants most often reported that they would turn to their spouses first.
Polarizing Effect on Relationships

We found that VHL can have a polarizing effect on relationships, increasing the closeness of some and the distance of others. This effect seemed to apply to different types of relationships, including familial, romantic, and friendships.

Overall, participants provided more examples of how VHL has brought them closer with others than how VHL created distance within relationships.

“I didn’t share as much personal stuff with [my mom] before, and now she is one of my best friends. I tell her everything.”—Seth

Impact on Life Decisions

Young adults often face major life decisions, and we found that VHL can have an impact on decisions regarding location, careers, and childbearing. Living with VHL impacted where two participants lived, influenced three participants to pursue careers in the healthcare field, and led to childbearing concerns for nine participants.

“I see the fun and the joy [a child] could bring. But a 50 percent chance of passing this VHL onto any kids I have is way too high of a percentage for me to really consider that.”—Scott

Assisted reproductive technology was discussed with the participants as a way to prevent having a child with VHL and half were open to utilizing in vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD), while two participants did not support the use of this technology. Two of the five participants who were open to IVF and PGD were not aware that these technologies existed until mentioned during the interview; neither of them had received genetic counseling in the past.

In conclusion, there is more to VHL than tumor development and surgeries. It leads to perpetual uncertainty, can affect relationships (in both positive and negative ways), and is often a consideration in life decisions. Those who fall within this young adult age group would likely benefit from establishing a long-term relationship with a genetic counselor. They can be the guide to finding ways to manage uncertainty, recommend VHL support groups, and provide information about available reproductive options.

The hope is that this study will spark the interest of researchers and provide inspiration for larger and more focused studies of this neglected age group, with the ultimate goal of providing more comprehensive care for VHL patients.

Note: Participants’ names have been changed to protect their privacy.