

The lived experience of having a rare medical disorder: Hermansky-Pudlak Syndrome

Linda D. Wagner, EdD, MSN, RN

Stacy Christensen, APRN, DNP, CCHP

Melissa Coleman, APRN, FNP-BC

Objectives

- 1. The learner will be able to verbalize the role of the nurse in working with individuals with a rare genetic disorder.
- 2. The learner will be able to verbalize four core themes related to the experience of living with Hermansky-Pudlak Syndrome.



Hermansky- Pudlak Syndrome

- Rare form of albinism
- Affects 1:500,000 to 1:1,000,000 non-Hispanics
- Affects 1:1800 individuals from Northwestern Puerto Rico
- Other symptoms include bleeding disorders, vision impairment, inflammatory bowel disease, pulmonary fibrosis

Qualitative Study

- Semi-structured face-to-face interviews
- HSC approved
- Setting: Annual National HPS Conference
- Sample: male or female, age 20-50, able to self report, speak English, identified diagnosis of HPS

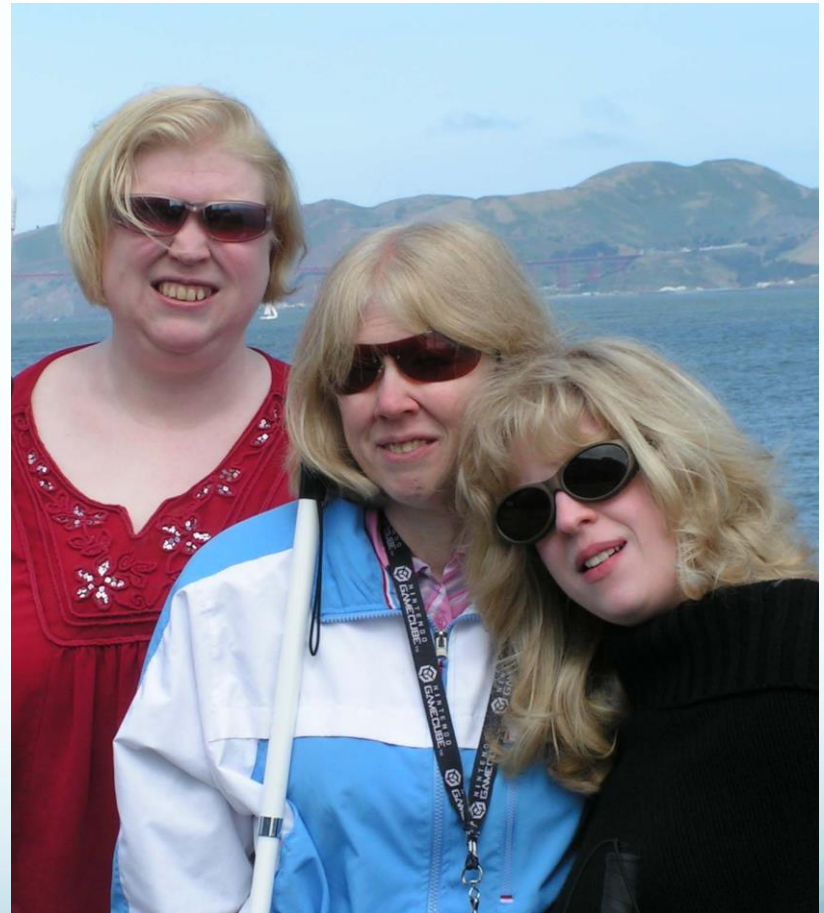
Sample

- Gender: M-4, F-19
- Marital status: Single-17, Married- 6
- Education: HS- 4, College-12, Beyond College- 7
- Age: current age-36.2, average age at diagnosis- 20.8



Findings

- Long road to diagnosis
- Giving, in order to move forward
- Survival is to belong to community
- Burden of being expert



Long Road to Diagnosis

- “The doctors did not know exactly what was going on and why I kept getting reoccurring infections. Having been sick from age 4 to age 13 and the doctors not knowing anything...they just diagnosed me with all kinds of different things.”
- “It has been very difficult to ever get a diagnosis. So a lot of things happened in my world that may never have needed to happen because I didn’t have an accurate diagnosis. So umm...I wasn’t diagnosed officially, it was a long journey, until I was 29.”

Giving, in order to move forward

- “My mom worried that I was obsessing too much and spending too much time in my life on my illness. And that wasn’t healthy. Whereas for me, it was the only way I could cope.”
- “I look at the clock and I can actually hear the hands of the seconds click each second of my life, and I’m running and I’m racing for my own life.”

Survival is to belong to community

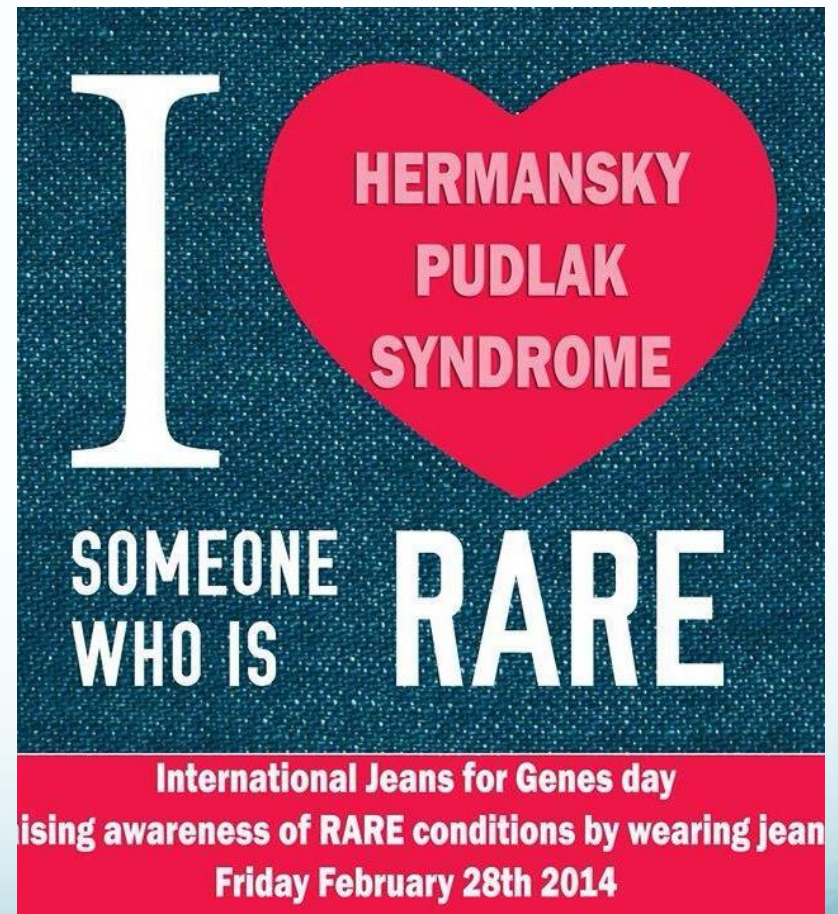
- “When you’re finding that commonality and you’re meeting a group of people that have the same disease as you do, it definitely doesn’t make it seem so rare anymore.”
- “Because there are so few of us, we all know each other and we all know each other’s parents and we all know each other’s siblings, and we all know each other’s cousins, and we all know each other’s husbands and wives and children, so then you know it becomes sort of like another family to come out to see.”

Burden of being expert

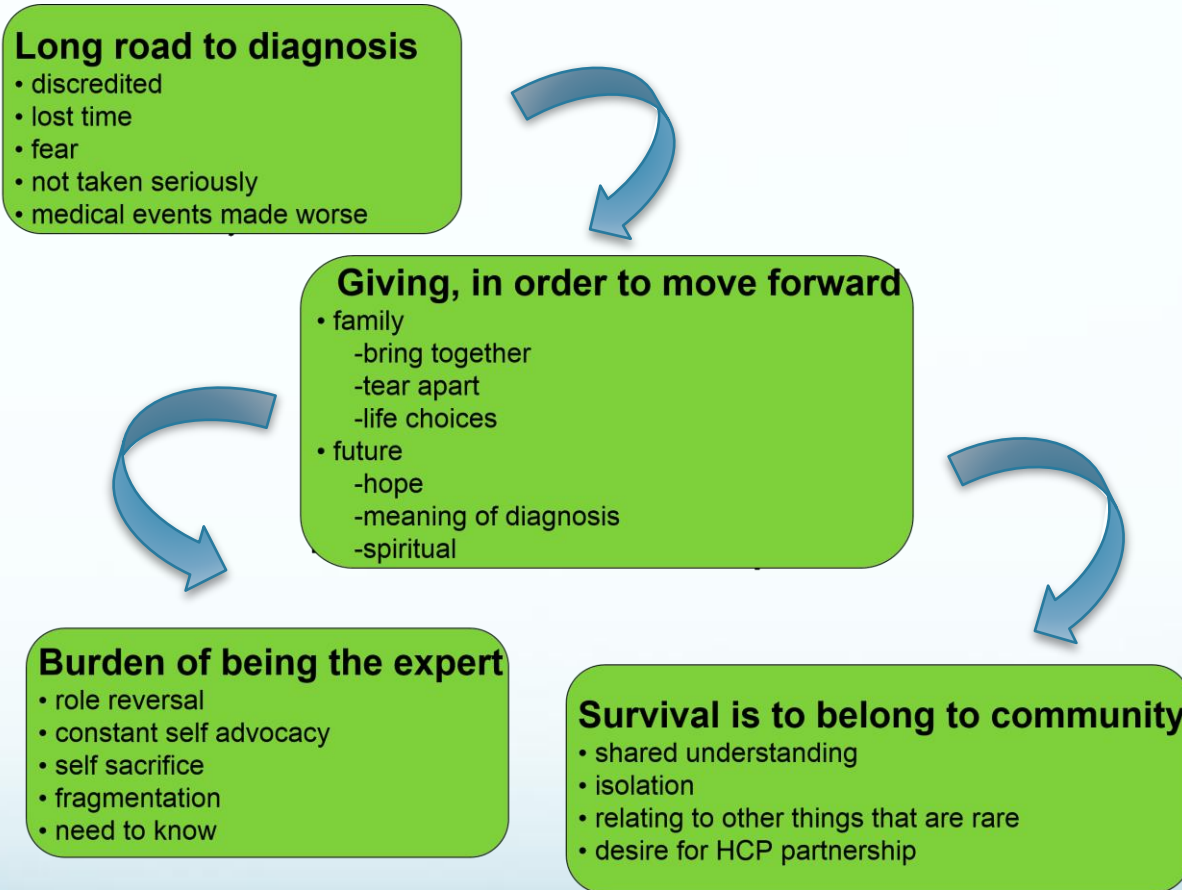
- “I’ve had to not only tell them what HPS is and what it does and spell it out and all that, but I’ve had to wait for them to pretend like they’re going to do something else because I’m blind they don’t think that I can see them far enough to see that they’re actually googling it on the computer. You know? Could you just flat out tell me you don’t know it?”
- “They don’t want to discuss things with you because you’re the patient; you’re not supposed to know things. But being able to articulate to them, ‘Listen I understand you’re the medical expert but when you went to medical school you never even heard of HPS. I’ve been listening to lectures on HPS for the last 2 years. I know a nationally known expert in this field. If you listen to me I can give you the basics.’ And so being your own personal advocate at that point is a big thing.”

Discussion

- Participants often feel discredited
- Role reversal with Practitioners
- HPS network vital to feeling less isolated



Model



Conclusion



Thank you

- The authors would like to thank Donna Appell for her support and assistance with the project, as well as all the individuals with HPS who enthusiastically volunteered their time to talk to us.



Questions