Living With WHIM Syndrome: The Patient Perspective

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BACKGROUND AND OBJECTIVES

- Warts, Hypogammaglobulinemia, Infections, Myelokathexis (WHIM) syndrome is a rare primary immunodeficiency caused by gain-of-function mutations in *CXCR4*, resulting in impaired immune cell trafficking and retention of neutrophils, lymphocytes, and monocytes in the bone marrow.[1,2]
- Recurrent bacterial, viral, fungal, and mycobacterial infections in people with WHIM syndrome may lead to substantial disease burden and life-threatening complications.[3-5]
- It is critical to understand the clinical journey of people with WHIM syndrome to identify challenges faced during the diagnostic process, allowing appropriate measures to be taken to aid in early and accurate diagnosis and treatment.
- This report explored the clinical journeys, experiences, and perceptions of people diagnosed with WHIM syndrome and/or their caregivers.

METHODS

- Interviews were conducted with people with WHIM syndrome and/or their caregivers in a group setting and individually to collect demographic and clinical information.
- All individuals completed a survey prior to the interview.
- Data obtained from the survey and interview were used to understand the clinical journey, experiences, and perceptions of these individuals living with WHIM syndrome.

RESULTS

3 individuals were recruited globally and interviewed

1 participant living with WHIM syndrome	1 participant living with WHIM syndrome and a caregiver	1 participant caring for a person with WHIM syndrome

EARLY SIGNS & SYMPTOMS

Participants reported a wide range of symptoms that varied over time, with recurrent infections leading to serious illnesses

Respiratory infections (upper respiratory tract)
Prompted first doctor's visit

Ear infections

Skin infections/warts

Bone infections

Missed myelokathexis
on bone marrow biopsy

PRE-DIAGNOSIS

Participants reported a long diagnostic journey, with visits to multiple HCPs, frequent misdiagnoses, and hospitalizations, with detrimental effects on psychological health

Frequent hospitalization

Misdiagnosis
Chronic neutropenia, CVID

Reduced quality of life
Impact on mental health, daily lives, and travel

Visits to immunologist, pediatrician, respiratory specialists, and hematologist

DIAGNOSIS

Genetic testing for *CXCR4* variants resulted in accurate diagnosis that helped reduce the psychological burden

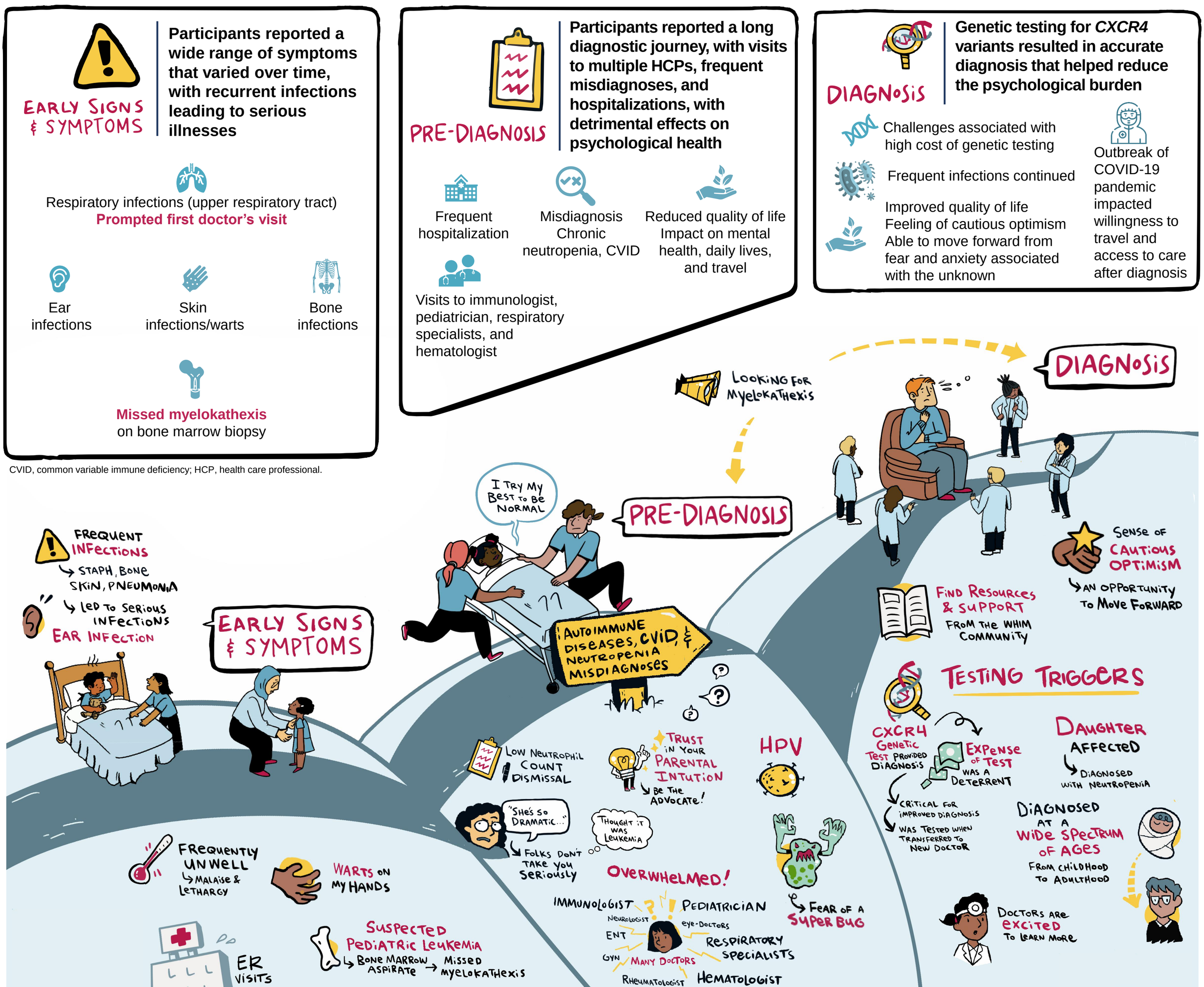
Challenges associated with high cost of genetic testing

Frequent infections continued

Improved quality of life
Feeling of cautious optimism
Able to move forward from fear and anxiety associated with the unknown

Outbreak of COVID-19 pandemic impacted willingness to travel and access to care after diagnosis

CVID, common variable immune deficiency; HCP, health care professional.



CONCLUSIONS

- **WHIM syndrome is a rare primary immunodeficiency with significant impact on the daily lives of people living with the disease.**
- **People living with WHIM syndrome and their caregivers reported a long, tiresome diagnostic journey, with frequent misdiagnoses associated with high psychological impact.**
- **Increased awareness and easy availability of no- or low-cost genetic testing for people with WHIM syndrome across a wide demography will help in early and accurate diagnosis of WHIM syndrome and provide opportunities for early therapeutic interventions.**
- **Outbreak of the COVID-19 pandemic impacted daily lives of the participants, reducing their willingness to travel for care or participate in different activities**
- **X4 Pharmaceuticals and Invitae have partnered to offer third-party–sponsored genetic testing and counseling through the PATH4WARD program at no charge for individuals who may carry a genetic mutation known to be associated with congenital or chronic neutropenia, including WHIM syndrome.**

References

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Disclosures

Michele Rhee is an employee and stock owner of X4 Pharmaceuticals.