PCR124

Jeffrey Thompson, PhD, MS¹; Bridget Balkaran, MPH¹; Doug Altenweg¹; Julie Flomenhoft¹; Amy Yanonis¹; Duygu Bozkaya² ¹Cerner Enviza, Malvern, PA, USA; ²Biogen, Cambridge, MA, USA

Introduction

group of rare eye disorders caused by an inherited gene mutation and can result in vision loss or blindness.¹ Some people with inherited retinal diseases, for instance those with retinitis pigmentosa (RP) or choroideremia (CHM), experience a gradual loss of vision, eventually leading to complete blindness.1-3

Resource Utilization

• Given the low prevalence, information is limited on the progression of these diseases, impact on quality of life, and healthcare resource utilization (HCRU).

Objective

• The objective of this study was to assess the impact of CHM on daily activities, work activities, and HCRU.

Methods

Study Design

impacted by CHM.

- Data was collected using a prospective, patient-reported survey fielded between December 2020 and October 2021.
- Eligible participants included males with a genetically confirmed CHM diagnosis receiving treatment by a clinician with a specialty in ophthalmology
- Participants were screened for eligibility and enrolled by one of eight participating physician-practices to complete an online survey assessing demographics, comorbidities, CHM-specific characteristics, use of a caregiver, HCRU, and activities
- For patients under age 18, the guardian/parent completed the patient interview.
- Inclusion criteria: Willing and able to provide informed consent.
 - Male diagnosed with CHM that has been confirmed via genetic testing.
- Patient has at least one record of visual acuity measurement in the past 24 months OR has be assigned by the treating clinician as having severe visual acuity impairment of blind/legally blind by using one of the following methods: finger counting, hand movement, light perception/no light perception.
- Exclusion criteria:

- Patients who received gene therapy or any other investigational treatment and patients who do not agree to informed consent.

Variables

- Demographic and clinical characteristics:
- Patient-reported information collected on: age, gender, race/ethnicity, employment, marital status, education, income, insurance type, and comorbidities (anxiety, depression, cancer, chronic kidney disease/renal failure, heart failure, hyperlipidemia, hypertension, obesity, osteoarthritis, type 2 diabetes, respiratory disease (e.g., asthma, COPD, emphysema), gastrointestinal disorders (e.g., IBD, ulcerative colitis, Crohn's), connective tissue disease (e.g., rheumatoid arthritis, scleroderma), neurological disorders (e.g., ALS, Alzheimer's), ear conditions/hearing loss, developmental delay/learning disabilities, other injuries and conditions due to external causes (e.g., cataracts, diabetic retinopathy, dry eye, glaucoma, macular disease).
- CHM-specific characteristics:
- Patient-reported information collected on: year first diagnosed, non-CHM eyerelated diagnoses prior to CHM diagnosis, time from symptom onset to physician
- diagnosis. Impact of CHM:
- Impact on employment and school
- Impact on productivity (measured via Work Productivity and Activity Impairment
- Impact on caregiving for others

Impact on family

- Impact on daily activities measured via:
- EuroQol-5D5 (EQ-5D, EQ-DY)
- Health Utilities Index⁶ (HUI3)
- WPAI

Methods, continued

HCRU:

- Medical/household aids used for CHM
- All-cause and CHM-related HCRU in the past 12 months including primary care physician visits, specialist visits, urgent care visits, emergency room visits, and
- Self-reported financial burden of CHM including annual out-of-pocket

Data Analysis

- Descriptive statistics were reported for all study variables:
- Mean. median. and standard deviation (SD) for continuous and discrete variables Frequencies and percentages for categorical variables

Results

- 47 patients participated in the survey, which was fielded between December 2020 through October 2021.
- The largest age groups were 40-49 (25.5%), 18-29 (19.1%), 30-39 (17.0%), and 65+ (14.9%)
- Most patients were white (n=35; 74.5%) and all had health insurance coverage.
- The majority of participants reported household income levels of over \$75,000 (44.7%) followed by \$50,000-\$75,000 (23.4%).
- The most commonly-reported comorbidities were cataracts (n=11, 23.4%), anxiety (n=8, 17.0%), and depression (n=7, 14.9%).
- 34.0% reported as having an other eye condition, with the most commonly selfreported other eye condition being Retinitis Pigmentosa (34.0%) followed by Gyrate
- The average number of years from symptom onset to CHM diagnosis was 9.6 (SD=12.6); the average number of years diagnosed with CHM was 18.7 (SD=17.5).

Impact of CHM on Daily/Work Activities

- The activity most reported as being affected by CHM was "participating in an activity you enjoy" (n=23, 48.9%) (**Table 2**).
- CHM affected job status, with 25.6% (n=11) of patients changing/leaving a job, 20.9% (n=9) retiring early, and 9.3% (n=4) working less or taking a leave of absence.

Impact of CHM on Daily/Work Activities: Validated Measures

- The mean EQ-5D summary score for the adult and pediatric CHM sample was 0.84 (SD:0.11) and 0.89 (SD:0.01), respectively (**Table 3**).
- Among working participants, the mean presenteeism and overall work impairment (WPAI) scores in the overall (N=22) sample was 12.73 (SD:22.29).
- The mean activity impairment (WPAI) score was 21.28 (SD:26.59).
- The mean HUI3 summary score 0.73 (SD:0.28).
- CHM affected job status, with 25.6% (n=11) of patients changing/leaving a job, 20.9% (n=9) retiring early, and 9.3% (n=4) working less or taking a leave of absence.

CHM-Related HCRU

- Amongst the total sample (N=47), 85.1% of patients self-reported as using at least one of the queried resources to help with vision (Table 4).
- The most common healthcare resources used to aid vision were corrective eyewear (n=35, 74.5%), extra lighting (n=31, 66.0%), and use of a cane or walking stick (n=18; 38.3%).
- Other reported resources used included 20- or 24-inch computer monitor/screen (27.7%), digital navigation aid/talking GPS (21.3%), magnifying glass (14.9%), extra-large keyboard (10.6%), reader pen/electronic reader/reader machine (8.5%), talking clock (8.5%), closet organizers (8.5%), assistive vision technology/digital technology (VR glasses) (4.3%), seeing eye dog (2.1%), and tracking devices for lost items (keys, wallet,
- When asking about CHM-related out-of-pocket financial burden, the most common response was "no burden" (40.4%), followed by "extremely high burden" (36.2%), "high burden" (21.3%), and "some burden" (2.1%).
- The most commonly-reported CHM-related HCRU used in the past 12 months was a
- specialist visit (61.7%) followed by primary care physician visit (12.8%).
- The most commonly-reported all-cause HCRU used in the past 12 months was a primary care physician visit (85.1%) followed by specialist visit (44.7%).

Results, continued

Table 1. Cohort Demographics and Comorbid Characteristics

| | Total Sample | |
|--|--------------|---------|
| Categorical Variables | N | % |
| Total | 47 | 100.0% |
| Age | <u> </u> | • 55 |
| 5-12 | 3 | 6.4% |
| 13-17 18-29 | <u> </u> | 6.4% |
| 30-39 | 8 | 17.0% |
| 40-49 | 12 | 25.5% |
| 50-64 | 5 | 10.6% |
| 65+ | 7 | 14.9% |
| Ethnicity/Race | | |
| African American/Black | 2 | 4.3% |
| Asian or Pacific Islander | 3 | 6.4% |
| Hispanic | 3 | 6.4% |
| Native American or Alaskan native | 1 | 2.1% |
| Other | 2 | 4.3% |
| White | 35 | 74.5% |
| Prefer not to say | l | 2.1% |
| No | 0 | 0.0% |
| Yes | 47 | 100.0% |
| Insurance Type | 17 | 100.070 |
| Private health insurance | 25 | 53.2% |
| Insurance purchased through a state exchange (Obamacare/ACA) | 2 | 4.3% |
| Medicare | 15 | 31.9% |
| Medicaid or other state plan | 2 | 4.3% |
| Other | 4 | 8.5% |
| Prescription Coverage | | |
| No | 3 | 6.4% |
| Yes | 44 | 93.6% |
| Marital Status | | |
| Single, never married | 11 | 23.4% |
| Committed relationship | 3 | 6.4% |
| Married | 24 | 51.1% |
| Separated | 2 | 4.3% |
| Divorced | 1 | 2.1% |
| Not applicable (under 18 years old) | 6 | 12.8% |
| Education | | 0.0% |
| Less than high school | 0 | 0.0% |
| Completed some high school High school graduate or equivalent (e.g., GED) | <u> </u> | 0.0% |
| Completed some college, but no degree | 9 | 12.8% |
| Associate's degree | 3 | 6.4% |
| College graduate (e.g., B.A., A.B., B.S.) | | 14.9% |
| Completed some graduate school, but no degree | 4 | 8.5% |
| Completed graduate school (e.g., M.S., M.D., Ph.D.) | 12 | 25.5% |
| Not applicable (under 18 years old) | 6 | 12.8% |
| Household Income | | 12.070 |
| Less than \$25,000 | 2 | 4.3% |
| \$25,000-\$49,000 | 4 | 8.5% |
| \$50,000-\$75,000 | 11 | 23.4% |
| Over \$75,000 | 21 | 44.7% |
| Prefer not to say | 3 | 6.4% |
| Not applicable (under 18 years old) | 6 | 12.8% |
| Other Eye Diagnoses | | |
| Retinitis Pigmentosa | 16 | 34.0% |
| RP Sine Pigmento | 0 | 0.0% |
| Stargart Disease | 0 | 0.0% |
| Usher Syndrome Type 1 | 0 | 0.0% |
| Kearns-Sayre Syndrome | 0 | 0.0% |
| Gyrate Atrophy | 2 | 4.3% |
| None of the above | 0 31 | 0.0% |
| Comorbidities | 31 | 66.0% |
| Anxiety | 8 | 17.0% |
| Depression | 7 | 14.9% |
| Chronic kidney disease/renal failure | 0 | 0.0% |
| Heart failure | 1 | 2.1% |
| Hyperlipidemia | 3 | 6.4% |
| Hypertension | 5 | 10.6% |
| Obesity | 5 | 10.6% |
| Osteoarthritis | 0 | 0.0% |
| Type 2 diabetes | 3 | 6.4% |
| Respiratory disease (e.g., asthma, COPD, emphysema) | 3 | 6.4% |
| Gastrointestinal disorders (e.g., IBD, ulcerative colitis, Crohn's) | 2 | 4.3% |
| Connective tissue disease (e.g., rheumatoid arthritis, scleroderma) | 0 | 0.0% |
| Neurological conditions (e.g., ALS, Alzheimer's) | 1 | 2.1% |
| Ear conditions/hearing loss | 6 | 12.8% |
| Developmental delay/learning disabilities | 4 | 8.5% |
| Other injuries and conditions due to external causes | 4 | 8.5% |
| Cataracts | 11 | 23.4% |
| Diabetic retinopathy | 0 | 0.0% |
| Dry eye | 5 | 10.6% |
| Glaucoma Macular disease | <u> </u> | 2.1% |
| Macular disease | 20 | 2.1% |
| None of the above | 20 | 42.6% |

Continuous Variables

Years from Symptoms to CHM Diagnosis

Years Diagnosed with CHM

CHM: Choroideremia; SD: standard deviation

Mean SD

Table 2. Impact of CHM on Daily Activities and Employment Status

| | | Total Sample | |
|--|----|--------------|--|
| Categorial Variables | N | % | |
| otal | 47 | 100.0% | |
| mpact of CHM | | | |
| Changed jobs or left a job | 11 | 23.4% | |
| Stopped attending school or changed school schedule | 3 | 6.4% | |
| Worked less hours or take a leave of absence from a job | 4 | 8.5% | |
| Retired early | 9 | 19.1% | |
| Stopped participating in an activity you enjoy | 23 | 48.9% | |
| Needed someone to accompany you to/from somewhere you received treatment | 19 | 40.4% | |
| Needed someone to care for you in a way they normally would not need to | 12 | 25.5% | |
| mployment Status: Current | | | |
| Employed full-time | 20 | 42.6% | |
| Employed part-time | 2 | 4.3% | |
| Self-employed | 1 | 2.1% | |
| Not employed, but looking for work | 2 | 4.3% | |
| Not employed and not looking for work | 3 | 6.4% | |
| Retired | 5 | 10.6% | |
| Short-term disability | 1 | 2.1% | |
| Long-term disability | 5 | 10.6% | |
| Student | 8 | 17.0% | |
| Homemaker | 0 | 0.0% | |
| imployment Status: Before CHM | | | |
| N Answered | 18 | 38.3% | |
| Employed full-time | 12 | 66.7% | |
| Employed part-time | 2 | 22.2% | |
| Self-employed | 4 | 11.1% | |
| Not employed, but looking for work | 0 | 0.0% | |
| Not employed and not looking for work | 0 | 0.0% | |
| Retired | 0 | 0.0% | |
| Short-term disability | 0 | 0.0% | |
| Long-term disability | 0 | 0.0% | |
| Student | 0 | 0.0% | |
| Homemaker | 0 | 0.0% | |

Table 3. Validated Measures

| | Mean | SD |
|--|-------|-------|
| EQ-5D Summary Score (HRQoL): Youth (<18) | 0.89 | 0.01 |
| EQ-5D Summary Score (HRQoL): Adult | 0.84 | 0.11 |
| Absenteeism (WPAI) | 0.00 | 0.00 |
| Presenteeism (WPAI) | 12.73 | 22.29 |
| Overall Work Impairment (WPAI) | 12.73 | 22.29 |
| Activity Impairment (WPAI) | 21.28 | 26.59 |
| HUI3 Summary Score | 0.73 | 0.28 |

Table 4. CHM-Related HCRU

| Categorical Variables | | Total Sample | |
|---|------|---------------------|--|
| | | % | |
| Total | 47 | 100.0 | |
| Resources Used to Help with Vision | | | |
| Corrective eyewear (glasses or lenses) | 35 | 74.5% | |
| Magnifying glass | 7 | 14.9% | |
| Cane or walking stick | 18 | 38.3% | |
| Assistive vision technology/digital technology (VR glasses) | 2 | 4.3% | |
| Seeing eye dog | 1 | 2.1% | |
| 20- or 24-inch computer monitor/screen | 13 | 27.7% | |
| Extra-large keyboard | 5 | 10.6% | |
| Reader pen/electronic reader/reader machine | 4 | 8.5% | |
| Talking clock | 4 | 8.5% | |
| Digital navigation aid/talking GPS | 10 | 21.3% | |
| Tracking devices for lost items (keys, wallet, etc.) | 1 | 2.1% | |
| Closet organizers | 4 | 8.5% | |
| Extra lighting | 31 | 66.0% | |
| Other | 7 | 14.9% | |
| None of the above | 7 | 14.9% | |
| Annual Out-of-Pocket Expenditures for CHM | | | |
| Haven't spent any money out of pocket | 8 | 17.0% | |
| Less than \$100 | 6 | 12.8% | |
| \$100-\$500 | 11 | 23.4% | |
| \$501-\$1,000 | 11 | 23.4% | |
| \$1,001-\$2,000 | 2 | 4.3% | |
| \$2,001-\$5,000 | 5 | 10.6% | |
| More than \$5,000 | 2 | 4.3% | |
| Don't know | 2 | 4.3% | |
| CHM-Related Out-of-Pocket Financial Burden | | | |
| No burden | 19 | 40.4% | |
| Extremely high burden | 17 | 36.2% | |
| High burden | 10 | 21.3% | |
| Moderate burden | 0 | 0.0% | |
| Some burden | 1 | 2.1% | |
| How Often Overwhelmed with CHM Out-of-Pocket Financial Burden | | | |
| Not at all | 21 | 44.7% | |
| Very often | 13 | 27.7% | |
| Often | 11 | 23.4% | |
| Somewhat often | 2 | 4.3% | |
| Rarely | 0 | 0.0% | |
| CHM-Related HCRU (Past 12 Months) | | | |
| Primary care physician | 6 | 12.8% | |
| Specialist | 29 | 61.7% | |
| Urgent care | 1 | 2.1% | |
| Emergency room visit | 0 | 0.0% | |
| Hospital (admitted/hospitalized) | 0 | 0.0% | |
| All-Cause HCRU (Past 12 Months) | | | |
| Primary care physician | 40 | 85.1% | |
| Specialist | 21 | 44.7% | |
| Urgent care | 9 | 19.1% | |
| Emergency room visit | 3 | 6.4% | |
| Hospital (admitted/hospitalized) | 8 | 17.0% | |
| Continuous Variables | Mean | SD | |
| | Mean | 30 | |
| CHM-Related HCRU (Past 12 Months) | 1 7 | 0.4 | |
| Primary care physician Specialist | 1.7 | 0.4 | |
| Specialist | 2.1 | 2.0 | |
| Urgent care | 1.0 | N/A | |
| Emergency room visit | _ | | |
| Hospital (admitted/hospitalized) | | | |
| All-Cause HCRU (Past 12 Months) | 2.2 | 7.0 | |
| Primary care physician | 2.0 | 1.2 | |
| Specialist | 3.5 | 3.4 | |
| Urgent care | 1 7 | 0.7 | |

HCRU: healthcare resource utilization; SD: standard deviation

Hospital (admitted/hospitalized)

Emergency room visit

Limitations

• The major limitation of this study is recall bias. Participants who completed the survey may have reported incomplete or inaccurate answers to the study questions, and thus may bias the analysis based on these answers.

Conclusions

- Choroideremia-related visual impairment is associated with negative impacts on both daily activities and work-related activities.
- Nearly half of all patients reported that CHM caused them to have "stopped participating in an activity you enjoy"; of this overall sample, 23.4% of participants reported that they "changed jobs or left a job" as a result of their CHM, 20.9% reported retiring early, and 9.3% reported working less or taking a leave of absence.
- The majority of patients with CHM (85.1%) do use other resources to help with vision. The most commonly used resource to help with vision was corrective eyewear (glasses or lenses) at 74.5%, followed by extra lighting, which was reported as being used by 66.0% of participants.
- Over 50% of CHM patients reported that the CHM-associated out-ofpocket costs represented an extremely high or high burden.

References

- Sahel J, Marazova S, et al. Clinical Characteristics and Current Therapies for Inherited Retinal Degenerations. Cold Spring Harb Perspect Med. doi: 10.1101/cshperspect.a017111.
- . National Institutes of Health. National Library of Medicine. Genetics Home Reference "Choroideremia." https://ghr.nlm.nih.gov/condition/choroideremia. Reviewed July 2013. Accessed October 10, 2018
- 3. Nash BM, Wright DC, et al. Retinal dystrophies, genomic applications in diagnosis and prospects for therapy. *Trans Pediatr.* 2015;4(2):139-163.
- 4. Reilly MC, Zbrozek AS, Dukes EM. The validity and reproducibility of a work productivity and activity impairment instrument. *Pharmacoeconomics*. 1993 Nov 1;4(5):353-65
- 5. Herdman M, Gudex C, Lloyd A, Janssen MF, Kind P, Parkin D, Bonsel G, Badia X. Development and preliminary testing of the new five-level version of EQ-5D (EQ-5D-5L). Quality of life research. 2011 Dec 1;20(10):1727-36.
- 6. Horsman J, Furlong W, Feeny D, Torrance G. "The Health Utilities Index (HUI®): Concepts, Measurement Properties and Applications." Health and Quality of Life Outcomes (electronic journal) (Vol. 1: 54, October 16, 2003), at http://www.hqlo.com/content/1/1/54.

Funding and Disclosures

- This study was funded by Biogen.
- Duygu Bozkaya is a former employee and stockholder of Biogen. Jeffrey Thompson, Bridget Balkaran, Doug Altenweg, Julie Flomenhoft, and Amy Yanonis are employees of Cerner Enviza (formerly Kantar Health), which received funding from Biogen to conduct this study.

Presented at ISPOR 2022, National Harbor, Maryland, USA, May 15-18, 2022