SUPPLEMENTARY MATERIAL

Quality of Life and Economic Impacts of Retinitis Pigmentosa on Japanese Patients: A Non-interventional Cross-sectional Study

Katsuhiko Watanabe [1], Celia Aouadj [2], Yoshimune Hiratsuka [3], Shuichi Yamamoto [4], Akira Murakami [3]

[1]Ophthalmology Medical Franchise Department, Novartis Pharma K.K., Tokyo, Japan

[2]HEOR Group, Novartis Pharma K.K., Tokyo, Japan

[3]Department of Ophthalmology, Juntendo University, Graduate School of Medicine, Tokyo, Japan

[4]Japan Community Health Care Organization, Tokyo, Japan

Corresponding author:

Katsuhiko Watanabe

Toranomon Hills Mori Tower 23-1, Toranomon 1-chome Minato-ku, Tokyo 105-6333, Japan

Phone: +81 80-3026-4262

Email: katsuhiko.watanabe@novartis.com

ORCID iD: 0000-0002-3060-0926

Contents

Supplementary Data S1 Questionnaire

Module 1. About you (the person affected by retinitis pigmentosa or LCA)

In this Module, you, the person affected by retinitis pigmentosa or LCA, will be asked about your personal information.
After a question text, answer options starting with alphabets are listed.
Please select what is applicable. When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

Q1. Are you;

a. Female
b. Male
c. Prefer not to say

Q2. What is your age?

a. years
b. Prefer not to say

Q3. Which of the options best describes your vision? Select all that apply.

a. I do not feel even light
b. I can see light or shadows
c. Some useful central vision
d. Good central vision
e. Some useful peripheral vision
f. Good peripheral vision
g. I experience night blindness
h. I still have good overall vision
i. Other – please specify

Q4. Which of the options best describes your mobility? Select only one that applies.

a. My vision doesn’t give any clue for me to walk through obstacles. I have to rely only on my cane, on sounds or on my guide dog or a companion person.
b. I use my cane for obstacle detection. I also use my remaining vision for additional support. I can see light differences or shadows that help me navigate.
c. My mobility is slower than before. I rely primarily on my vision. I might rely on a cane for additional support.

d. My mobility is almost normal but I have to scan my environment for potential obstacles. I do not need to use a cane.

e. My mobility is normal but occasionally I bump into obstacles. Sometimes I am surprised by events on my side.

Q5. Which of the options best describes your vision when you read books, etc. Select only one that applies. If you are wearing your glasses or contact lenses, answer when you are wearing them.

a. I do not see anything or I can only see light but no movement.

b. I can see light, shadows, and movement. I cannot read using my vision. I rely on talking books, braille or other nonvisual sources.

c. I may be able to read with vision aids but I prefer talking books.

d. I can read with vision aids but slower than before. I use high power magnifiers.

e. I have an almost normal reading capacity. I just bring the text closer to my eyes for reading. But I can read small print. I use low power magnifiers lens or large print reading books.

f. I have no issues with reading, even with small print. My reading speed is normal. I read at normal distance.

Q6. What is your level of vision and visual acuity? Answer that with glasses or contact lenses, if you wear them.

a. Enter your visual acuity in number:

b. Counting finger (being able to count the number of fingers held in front of you)

c. Hand motion (being able to see the hand move in front of your eyes)

d. Light perception (being able to feel light)

e. No light perception (do not even feel light)

f. Unsure

Q7. Do you have retinitis pigmentosa or Leber congenital amaurosis (LCA)? (Check with your primary doctor as needed.)

a. Retinitis pigmentosa

b. LCA

Q8. What kind of retinitis pigmentosa do you have? (Check with your primary doctor as needed.)

a. Autosomal dominant inherited type

b. Autosomal recessive inherited type

c. X-linked inherited type (X-linked recessive inherited type)

d. Sporadic type (None of your relatives has the same disease.)

e. Don’t know

Q9. What kind of LCA do you have? (Check with your primary doctor as needed.)

a. Autosomal dominant inherited type

b. Autosomal recessive inherited type

c. Sporadic type (None of your relatives has the same disease.)

d. Don’t know

Q10. Have you received a genetic testing specifically for your inherited retinal disease? (Check with your primary doctor as needed.)

a. Yes

b. No

c. Don’t know

Q11. What gene was identified to be causing your condition?

a. Please specify the identified gene:

b. Unsure

c. No gene identified

d. Gene identified but prefer not to say

Q12. Do you have a physical disability certificate?

a. Yes (Certified class: Class)

b. No – Currently pending

c. No – Applied but not issued

d. No – Not applied

Q13. What is the reason why you did not apply for a physical disability certificate? Select all that apply.

a. The procedure is troublesome

b. I do not feel merit for possessing it or feel demerit

c. Other – please specify

Q14. Are you a student now?

a. Yes
b. No

Q15. Do you have a part-time job now?

a. Yes
b. No

Q16. What is the highest level of formal education you have completed to date? (If you are a student now, select the last school you graduated from.)

a. Primary school
b. Junior high school
c. Senior high school
d. Bachelor’s degree
e. Master’s degree
f. Doctoral degree
g. Other – please specify:
h. Prefer not to say

Module 2 Work

In this Module, you will be asked about your work.
After a question text, select an answer or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.
If you are currently a student, read the word “work” in question texts as “part-time job,” and answer the questions.

The following questions will ask you in detail about different aspects of your employment and how your health problems may have impacted your work. Moreover, to learn about your usual situation properly, you will be asked how the spread of COVID-19 infections impacted your current situation.

Q1 to Q6. Work Productivity and Activity Impairment Questionnaire: General Health, Version 2.0 (WPAI:GH V2.0) (Home (reillyassociates.net))

Q7. Has the spread of COVID-19 infections affected your work or employment status?

a. Yes

b. No

Q8. During the past 7 days, how many hours has the spread of COVID-19 infections impacted the time you missed from work because of your health problems? (If 0 hours, please enter the number “0 [zero].”)
Answer: hour(s)

Q9. During the past 7 days, how many hours has the spread of COVID-19 infections impacted the time you missed from work because of any reason other than your health problems? (If 0 hours, please enter the number “0 [zero].”)
Answer: hour(s)

Q10. How many hours do you usually work per week?
Answer: hour(s)

Q11. During the last 1 year, how many full and partial days (e.g., 1.5 days) did you miss from work because of any other reason, including refreshment and illness? (If 0 days, please enter the number “0 [zero].”)
Answer: day(s)

Q12. During the last 1 year, how many full and partial days (e.g., 1.5 days) did you miss from work due to your visual impairment (including hospital visits, hospitalization, and surgery)? (If 0 days, please enter the number “0 [zero].”)
Answer: day(s)

Q13. How does your visual impairment impact your job performance over a regular 4-week period? Select from the following 6 scales from 0 to 5.

0. No impact	1. Mild impact	2. Mild to moderate impact	3. Moderate impact	4. Moderate to significant impact	5. Significant impact
○	○	○	○	○	○

Q14. Please select any articles for daily use that you use to support your vision in your working environment. Select all that apply.

a. Computers (select only if specialized for you)

b. Magnifying glasses (magnifier)

c. Magnification apparatus for reading

d. Screen reader, spoken-word processor, or verbal calculator

e. Braille display, braille books, braille device

f. Modifications to mobile phones, including smart phones (applications, etc.)

g. Digital voice recorder (voice reading machine)

h. Not using any items

i. Other – please specify:

Q15. How much money do you spend on average per year for articles for daily use to support your vision in your working environment? Please specify the amount of money you paid and, if you know, the amount of subsidies. (If 0 yen, please enter the number “0 [zero].”)

- Amount of money you pay ○a. Cannot answer ○b. Can answer – Enter the amount: yen
- Amount of public subsidies ○a. Cannot answer ○b. Can answer – Enter the amount: yen
- Amount of subsidies from the employer ○a. Cannot answer ○b. Can answer – Enter the amount: yen

Q16. Has the spread of COVID-19 infections changed how much money you spend per year on articles for daily use to support your vision in your working environment?

- Amount of money you pay ○a. Increased – Enter the amount increased: yen ○b. Decreased – Enter the amount decreased: yen ○c. Unchanged
- Amount of public subsidies ○a. Increased – Enter the amount increased: yen ○b. Decreased – Enter the amount decreased: yen ○c. Unchanged

- Amount of subsidies from the employer ○a. Increased – Enter the amount increased: yen ○b. Decreased – Enter the amount decreased: yen ○c. Unchanged

Q17. Please select the most appropriate reason for not currently using articles for daily use to support your vision in your working environment.

a. Do not require articles for daily use to support my vision
b. Cannot use articles for daily use to support my vision
c. Other – please specify:

Q18. Which of the following best describes your current state of work?

a. I work as a full-time worker
b. I work regularly as a part-time worker
c. Self-employed
d. Casual work (not working on a regular basis)
e. Other – please specify:

Q19. Which of the following best describes your current state of work?

a. Vocational training (not working)
b. Retired
c. Not employed (looking for work)
d. Not employed (not looking for work but working around the house, childcare, providing care, etc.)
e. Unable to work
f. Other – please specify:

Q20. How many years have you worked at the current place of employment?
Answer: year(s)

Q21. Please describe your current occupation.
Answer: Complete freely.

Q22. What was the annual income you earned from working in the last year (total annual income before tax and insurance fee)?

a. yen
b. Prefer not to say

Q23. Have you ever been refused at a job interview due to your visual impairment?

a. Yes

b. No

c. Never had a job interview

Q24. For what type of work were you refused at a job interview due to your visual impairment? Please describe concretely.

- Prefer not to say
- Column for providing the answer:

Q25. Have you ever given up your work due to your visual impairment? Or, have you ever failed to have a work you wanted?

a. Yes

b. No

Q26. What kind of work you had to give up? Or what kind of work you could not have? Please describe concretely.

- Prefer not to say
- Column for providing the answer:

Module 3 Education

In this Module, you will be asked about your education.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

Q1. Which of the following describes the type of education that you receive now?

a. General school or education without special teaching methods or support for visual impairment
b. School or education with special classes for handicapped students
c. School or education with special support classes
d. Special support school for the visually impaired
e. Special support school for handicapped students other than the visually impaired
f. Other – please specify:

Q2. How many hours per week altogether do you have classes in a curriculum? (If 0 hours, please enter the number “0 [zero].”)

Answer: hour(s)

Q3. Usually, how many extra hours per week altogether do you study at your place of learning (school and tutoring school) other than the time of classes specified in the curriculum? (If 0 hours, please enter the number “0 [zero].”)

Answer: hour(s)

Q4. Usually, how many hours per week do you spend on education-related activities (e.g., studying, completing homework and/or assignments, research etc.) outside of your place of learning (at home, etc.)? (If 0 hours, please enter the number “0 [zero].”)

Answer: hour(s)

Q5. During the past 1 year, how many full and partial days of class (e.g., 1.5 days) did you miss? (If 0 days, please enter the number “0 [zero].”)

Answer: day(s)

Q6. During the past 1 year, how many full and partial days of class (e.g., 1.5 days) did you miss because of your visual impairment (including hospital visits, hospitalization, surgery, etc.)? (If 0 days, please enter the number “0 [zero].”)

Answer: day(s)

Q7. During the past 7 days, how much visual impairment affected your study?

Think about days you were limited in the amount or kind of study you could do, days you accomplished less than you would like, or days you could not do your study as carefully as usual. If your visual impairment affected your study only a little, choose a low number. Choose a high number if it affected your study a great deal.

Consider only how much your visual impairment affected studying.

Visual impairment had no effect on my study	○ 0	○ 1	○ 2	○ 3	○ 4	○ 5	○ 6	○ 7	○ 8	○ 9	○ 10	Visual impairment completely prevented me from studying

Q8. During the past 7 days, how much did your visual impairment affect your regular daily activities, other than studying?

The regular activities refer to the usual activities you do, such as work around the house, shopping, childcare, exercising, etc. Think about times you were limited in the amount or kind of activities you could do and times you accomplished less than you would like. If your visual impairment affected your activities only a little, choose a low number. Choose a high number if it affected your activities a great deal.

Consider only how much your visual impairment affected your regular daily activities, other than studying.

Visual impairment had no effect on my daily activities	○ 0	○ 1	○ 2	○ 3	○ 4	○ 5	○ 6	○ 7	○ 8	○ 9	○ 10	Visual impairment completely prevented me from doing my daily activities

Q9. Please select any articles for daily use that you are currently using to support your vision in your learning environment. Select all that apply.

a. Computers (select only if specialized for you)

b. Magnifying glasses (magnifier)

c. Magnification apparatus for reading

d. Screen reader, spoken-word processor, or verbal calculator

e. Braille display, braille books, braille device

f. Modifications to mobile phones, including smart phones (applications, etc.)

g. Digital voice recorder (voice reading machine)

h. Not using any items

i. Other – please specify:

Q10. How much money do you spend on average per year for articles for daily use to support your vision in your learning environment? Please specify the amount of money you or your family paid and, if you know, the amount of subsidies. (If 0 yen, please enter the number “0 [zero].”)

- Amount of money you or your family paid ○a. Cannot answer ○b. Can answer – Enter the amount: yen
- Amount of public subsidies ○a. Cannot answer ○b. Can answer – Enter the amount: yen

Q11. Has the spread of COVID-19 infections changed how much money you spend per year on articles for daily use to support your vision in your learning environment?

- Amount of money you or your family paid ○a. Increased – Enter the amount increased: yen ○b. Decreased – Enter the amount decreased: yen ○c. Unchanged
- Amount of public subsidies ○a. Increased – Enter the amount increased: yen ○b. Decreased – Enter the amount decreased: yen ○c. Unchanged

Q12. Please select the most appropriate reason for not currently using articles for daily use to support your vision in your learning environment.

a. Do not require articles for daily use to support my vision

b. Cannot use articles for daily use to support my vision

c. Other – please specify:

Q13. Do you receive support for your learning from someone? Select all that apply.

a. Paid caregiver

b. Family members

c. Friends or classmates
d. Teachers
e. Volunteers
f. Not receiving support from anybody
g. Other – please specify:

Q14. Did you have to give up academic learning due to your visual impairment?

a. Yes
b. No

Q15. What academic learning did you have to give up? Please describe concretely.

- Prefer not to say
- Column for providing the answer:

Module 4 Healthcare services for retinitis pigmentosa or LCA

In this Module, you will be asked about healthcare services (hospital visits, etc.) for retinitis pigmentosa or LCA.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

Q1. Which health providers do you see for the management of your retinitis pigmentosa or LCA? Select all that apply.

a. Ophthalmologist (general)
b. Ophthalmologist (specializing in retinitis pigmentosa)
c. Pediatrician
d. Orthoptist, instructor for daily living training for the visually impaired (walking training instructor, etc.)
e. Psychologist, psychiatrist, or counselor
f. Genetic counselor
g. None
h. Other – please specify

Q2. How often do you attend appointments with each healthcare practitioner specifically for your retinitis pigmentosa or LCA?

a. Once a week
b. Once every 2 weeks
c. Once a month
d. Once every 3 to 6 months
e. Once a year
f. Once every 2 years
g. Other – please specify

Q3. Approximately how much money did your last visit to each healthcare practitioner cost you or your family out of pocket? (If 0 yen, please enter the number “0 [zero].”)

( ) yen

Q4. Do you take time off work or education to attend medical appointments?

a. Yes, for all of my appointments

b. Yes, for about three quarters of my appointments
c. Yes, for about half of my appointments
d. Yes, for about one quarter of my appointments
e. No, I do not take time off work or education to attend my appointments

Q5. Did you attend your medical appointments alone or with a caregiver? Select the highest frequency among the options.

a. I attend appointments alone
b. I attend appointments with an unpaid caregiver (family member or friend)
c. I attend appointments with a paid caregiver (paid to provide care for a patient with a visual impairment)
d. I attend appointments with an unpaid caregiver (family member or friend) and paid caregiver

Q6. Did your unpaid caregiver take time off work or education to attend these medical appointments?

a. Yes, for all of my appointments
b. Yes, for about three quarters of my appointments
c. Yes, for about half of my appointments
d. Yes, for about one quarter of my appointments
e. No, he/she did not take time off work or education to attend my appointments

Q7. Has the spread of COVID-19 infections reduced the number of visits to healthcare providers?

a. Yes
b. No

Q8. Approximately how long does it take for you to travel one way to your doctor/clinician for the management of your retinitis pigmentosa or LCA?

a. Less than 15 minutes
b. Between 15 minutes and less than 30 minutes
c. Between 30 minutes and less than 1 hour
d. Between 1 hour and less than 4 hours
e. Between 4 hours and less than 8 hours
f. Between 8 hours and less than 12 hours

g. Between 12 hours and less than 24 hours

h. Over 1 day

Q9. Approximately how much did your last one-way travel to your doctor/clinician cost you or your family out of pocket? (If 0 yen, please enter the number “0 [zero].”)

One-way travel: yen

Module 5 Medication and supplements

In this Module, you will be asked about medication and supplements.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

Q1. Do you use any medications for your retinitis pigmentosa or LCA? (except for vitamins or nutritional supplements)

a. Yes

b. No

Q2. What medications do you use to manage your retinitis pigmentosa or LCA? Please separate any medications using commas.

Column for entering the names of medications:

Q3. Do you use any vitamins or nutritional supplements for your retinitis pigmentosa or LCA?

a. Yes

b. No

Q4. What vitamins or nutritional supplements do you use for your retinitis pigmentosa or LCA? Select all that apply.

a. Lutein

b. Omega-3 fatty acids

c. Vitamin A

d. Retinol palmitate

e. Vitamin B1

f. Vitamin C

g. Zeaxanthin

h. Zinc

i. Other – please specify:

Q5. How much money in total have you spent on medications, vitamins, and supplements specifically for your retinitis pigmentosa or LCA over the last month? (Please answer only costs you or your family paid out of pocket without including rebates and subsidies.) (If 0 yen, please enter the number “0 [zero].”)

Answer: yen

Module 6 Vision aids

In this Module, you will be asked about vision aids.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

Q1. Please select visual aids you have purchased specifically for your visual impairment in the last year. Select all that apply.

a. Prescription glasses or contact lenses
b. Magnifying glasses (magnifier)
c. Light-protecting lenses, sunglasses
d. Magnification apparatus for reading
e. I have not purchased any visual aids
f. Other – please specify:

Q2. Approximately how much money have you or your family members spent on visual aids you have purchased specifically for your visual impairment in the last year? (If 0 yen, please enter the number “0 [zero].”)

a. Prescription glasses or contact lenses: yen
b. Magnifying glasses (magnifier): yen
c. Light-protecting lenses, sunglasses: yen
d. Magnification apparatus for reading: yen
e. Other – Present the item(s) in Q1 again: yen

Q3. Are you renting a guide dog?

a. Yes
b. No

Q4. Approximately how much money have you or your family member spent on your guide dog for food, equipment, veterinary fees, and other miscellaneous expenses in the last year? (If 0 yen, please enter the number “0 [zero].”)
Answer: yen

Q5. Do you use a white cane for the visually impaired?

a. Yes
b. No

Q6. Approximately how much money have you or your family members spent on your white cane in the last year? (If 0 yen, please enter the number “0 [zero].”)
Answer: yen

Q7. Do you use the aid of a mini guide (voice guide) or other electronic mobility devices (GPS or sensor devices)?

a. Yes
b. No

Q8. Approximately how much money have you or your family members spent on this electronic mobility device? (If 0 yen, please enter the number “0 [zero].”)
Answer: yen

Q9. Which of the following modifications have you installed in your home specifically for your visual impairment? Select all that apply.

a. Customized bathtub(s)
b. Customized door frame(s)
c. Customized light
d. Customized stairs
e. Other – please specify:
f. I have not installed any home modifications (exclusive answer)

Q10. Specify the number and approximately how much money you or your family members have spent on the installation of the home modifications specifically for your visual impairment. Please state the overall costs rather than the cost per unit/item. (If 0 yen, please enter the number “0 [zero].”)

a. Customized bathtub(s) Number: , Total amount: yen
b. Customized door frame(s) Number: , Total amount: yen
c. Customized light Number: , Total amount: yen
d. Customized stairs Number: , Total amount: yen
e. Other – Present the item(s) in Q1 again: Number: , Total amount: yen

Q11. Do you use functions or applications on cell phone or smart phone specifically to support your visual impairment?

a. Yes
b. No

Q12. Which of the following functions do you use on your cell phone or smart phone specifically to support your visual impairment? Select all that apply.

a. Enlarging text
b. Screen reading software
c. Voice dictation
d. Braille inputs or displays
e. Navigation applications
f. Audiobooks
g. Talking camera applications (Seeing AI, etc.)
h. Community support applications (Be My Eyes, etc.)
i. Other – please specify

Q13. Approximately how much money have you or your family members spent on functions or applications on cell phone or smart phone specifically to support your visual impairment? (If 0 yen, please enter the number “0 [zero].”)
Answer: yen

Q14. Which of the following aids do you use at home specifically for your visual impairment? Select all that apply.

a. Magnifying glasses (magnifier)
b. High-intensity lamps
c. Book alternatives (large print or audiobooks)
d. Handheld magnifiers
e. Tactile or large-print labels
f. Large keyboards
g. Screen magnification technology
h. Screen reading software
i. Portable note takers such as a digital voice recorder
j. Customized clocks, watches, or timers
k. Other – please specify up to 3 items: ( )( )( )

l. I do not use any (exclusive answer)

Q15. Specify the number and approximately how much money you or your family members have spent on aids you use at home specifically for your visual impairment? Please state the overall costs rather than the cost per unit/item. (If 0 yen, please enter

the number “0 [zero].”)

a. Magnifying glasses (magnifier) Number: , Total amount: yen
b. High-intensity lamps Number: , Total amount: yen
c. Book alternatives (large print or audiobooks) Number: , Total amount: yen
d. Handheld magnifiers Number: , Total amount: yen
e. Tactile or large-print labels Number: , Total amount: yen
f. Large keyboards Number: , Total amount: yen
g. Screen magnification technology Number: , Total amount: yen
h. Screen reading software Number: , Total amount: yen
i. Portable note takers such as a digital voice recorder Number: , Total amount: yen
j. Customized clocks, watches, or timers Number: , Total amount: yen

k1. Other – Present Q14k1 again: Number: , Total amount: yen
k2. Other – Present Q14k2 again: Number: , Total amount: yen
k3. Other – Present Q14k3 again: Number: , Total amount: yen

Q16. Overall, in the past year, approximately how much out-of-pocket expenditure have you or your family member spent on items related to your visual impairment to assist you with your daily living? (If 0 yen, please enter the number “0 [zero].”)
Answer: yen

Q17. Taking into consideration all expenses that were previously mentioned, is there any other item which you or your family members pay for to help with your visual impairment? If “Yes,” please specify the item and approximate associated cost.

a. Yes – please specify the name(s) of item(s): Amount paid out of pocket: yen
b. No

Module 7 Paid care

In this Module, you will be asked about your paid care.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

The following questions will ask you in detail about paid care arrangements you may use to assist you with your activities of daily living.

Q1. Do you have a paid caregiver (a person who receives rewards or salaries to give care) to assist you with your activities of daily living specifically for your visual impairment?

a. Yes
b. No

Q2. Approximately how many hours per week do you receive assistance from a paid caregiver specifically for your visual impairment?

a. Less than 5 hours
b. 6 to 10 hours
c. 11 to 20 hours
d. 21 to 30 hours
e. 31 to 40 hours
f. Over 40 hours

Q3. On average, what cost does your paid caregiver charge per hour? Please specify the amount of money you or your family paid and, if you know, the amount including subsidies. (If 0 yen, please enter the number “0 [zero].”)

- Amount of money you or your family paid ○a. Cannot answer ○b. Can answer
  – Enter the amount: yen
- Amount of money including public subsidies ○a. Cannot answer ○b. Can answer – Enter the amount: yen

Q4. How was the cost of your paid caregiver covered? Select all that apply.

a. Myself out of pocket

b. Friend and family out of pocket
c. Publicly funded healthcare plan
d. Publicly funded social care plan
e. Private health insurance plan
f. Charitable organization
g. Other – please specify:

Module 8 Unpaid care

In this Module, you will be asked about your unpaid care.
After a question text, answer options starting with alphabets are listed.
Please select what is applicable.
Questions may be skipped depending on the answers.

The following questions will ask you in detail about unpaid care arrangements you may use to assist you with your activities of daily living.

Q1. Do you receive any assistance from an unpaid caregiver? An unpaid caregiver includes any person, such as a family member, friend, or neighbor, who is giving regular, ongoing assistance to another person without payment for the care given.

a. Yes
b. No

Q2. Please identify your primary unpaid caregiver. Select the person who gives you care for the longest time.

a. Spouse
b. Parent
c. Child
d. Other relatives
e. Friend
f. Other – please specify:

Q3. Approximately how many hours per week did you receive assistance from an unpaid caregiver?

a. Less than 5 hours
b. 6 to 10 hours
c. 11 to 20 hours
d. 21 to 30 hours
e. 31 to 40 hours
f. Over 40 hours

Module 9 Financial aid

In this Module, you will be asked about financial aid for visual impairment.
After a question text, select an answer starting with alphabets or enter a numerical value.
When you need to enter numbers in an answer, use one-byte characters.
Questions may be skipped depending on the answers.

The following questions will ask you in detail about financial aid you may receive specifically for your visual impairment.

Q1. Do you currently receive any financial aid such as public donation (charity), disability pension, public funds, and private funds, specifically for your visual impairment?

a. Yes
b. No

Q2. If you receive any financial aid, which of the following forms of assistance do you receive? Select all that apply. Also, how much money do you receive per month?

a. Public donation (charity): yen
b. Disability pension: yen
c. Government-funded healthcare (provision of medical care, e.g., prevention, diagnosis, or treatment for illness): yen
d. Government-funded social care (support with activities of daily living, e.g., feeding, washing, and dressing): yen
e. Privately funded health care (provision of medical care): yen
f. Privately funded social care (support with activities of daily living): yen
g. Money from friends and family: yen
h. Other – please specify: : yen

Module 10 Your well-being

In this Module, you will be asked about your well-being.
After a question text, answer options starting with alphabets are listed.
Please select what is applicable.

The following questions ask you about your well-being in relation to your visual impairment. After each question, please choose the response that best describes your situation.

Q1. Have you ever felt depressed due to your visual impairment?

a. Yes

b. No

Q2. In what situation did you feel depressed? Please describe concretely.

- Prefer not to say
- Column for providing the answer:

Q3. Have you ever felt anxious due to your visual impairment?

a. Yes

b. No

Q4. In what situation did you feel anxious? Please describe concretely.

- Prefer not to say
- Column for providing the answer:

Q5. Are you taking medications to manage feelings of depression or anxiety due to your visual impairment?

a. Yes

b. No

Module 11 Visual Function Questionnaire

In this Module, you will be asked about your visual function.
After a question text, answer options with numbers at the end are listed.
Please select what is applicable.

National Eye Institute Visual Functioning Questionnaire - 25 (Questions are copyrighted and not listed.)

Module 12 Health Utilities Index® (HUI®)

In this Module, you will be asked about the Health Utilities Index®.
After a question text, answer options starting with alphabets are listed.
Please select what is applicable.

Questions are copyrighted and not listed.

Module 13 Your today’s health condition

In this Module, you will be asked about your today’s health condition.
After a question text, select an answer.
Please select what is applicable.

The 5-level EQ-5D version (Questions are copyrighted and not listed.)

Supplementary Data S2 Retinitis pigmentosa diagnosis criteria

The following diagnosis criteria for retinitis pigmentosa were translated into English and adapted from the Japanese clinical practice guidelines for retinitis pigmentosa [1].

Subjective symptoms

1. Night blindness
2. Tunnel vision
3. Reduction in visual acuity
4. Photophobia (or hemeralopia)

Laboratory findings

1. Funduscopic findings (attenuation of retinal vessels, coarse retinal appearance, bone spicule pigmentation, multiple white spots, optic atrophy, and macular degeneration).
2. Electroretinographic abnormalities (attenuation type, negative type, disappearance type).
3. Hyper- or hypo-fluorescence caused by retinal pigment epithelium atrophy revealed by fundus autofluorescence testing.
4. Ellipsoid zone (inner/outer segment) abnormality of the fovea (non-continuity or disappearance) when examined by optical coherence tomography.

Diagnostic judgment

1. The disease is progressive.
2. At least one of the subjective symptoms listed above is present.
3. At least two of the fundus features listed above are visible.
4. The above-mentioned electroretinographic features are visible.
5. The disease is not inflammatory or secondary in nature.

If all five of the above criteria are present, a diagnosis of retinitis pigmentosa (one of the designated intractable diseases) is made.

Severity grading[a]

Cases of severity grades II, III, and IV are covered.

Grade I: Corrected visual acuity ≥0.7 and free of tunnel vision

Grade II: Corrected visual acuity ≥0.7 and tunnel vision present

Grade III: Corrected visual acuity <0.7 and ≥0.2

Grade IV: Corrected visual acuity <0.2

[a] Both the corrected visual acuity and the visual field adopt the value of the better side. “Tunnel vision present” means cases where the central residual visual field is within 20 degrees with the Goldmann I-4 optotypes.

Reference

1. Clinical practice guidelines for retinitis pigmentosa. Nippon Ganka Gakkai Zasshi. 2016;120:846–61 [in Japanese].

Supplementary Data S3 Criteria for the American Medical Association (AMA) visual impairment class

Corresponding response alternatives were prepared for the estimated visual ability by visual field and visual acuity impairment class described in AMA Guides to the Evaluation of Permanent Impairment.

The AMA visual impairment class for each patient was determined as follows:

1. The visual field impairment class was determined from the response to Question (Q) 4 in Module 1 and according to Table S3.1.
2. The visual acuity impairment class was determined from the response to Q5 in Module 1 and according to Table S3.2.
3. When a patient's responses matched two different impairment classes, the more severe impairment class was selected as the visual impairment class.

Examples:

- If a patient answered "a" to Q4 and "a" to Q5, the visual impairment class of the patient was determined as "Total Blindness".
- If a patient answered "d" to Q4 and "c" to Q5, the visual impairment class of the patient was determined as "Profound Low Vision".

Table S3.1 Visual field impairment class

Range of Normal Vision	–
Near-Normal Vision	e. My mobility is normal but occasionally I bump into obstacles. Sometimes I am surprised by events on my side.
Moderate Low Vision	d. My mobility is almost normal but I have to scan my environment for potential obstacles. I do not need to use a cane.
Severe Low Vision	c. My mobility is slower than before. I rely primarily on my vision. I might rely on a cane for additional support.
Profound Low Vision	b. I use my cane for obstacle detection. I also use my remaining vision for additional support. I can see light

	differences or shadows that help me navigate.
Near-Blindness	a. My vision doesn’t give any clue for me to walk through obstacles. I have to rely only on my cane, on sounds or on my guide dog or a companion person.
Total Blindness	–

Table S3.2 Visual acuity impairment class

Range of Normal Vision	f. I have no issues with reading, even with small print. My reading speed is normal. I read at normal distance.
Near-Normal Vision	–
Moderate Low Vision	e. I have an almost normal reading capacity. I just bring the text closer to my eyes for reading. But I can read small print. I use low power magnifiers lens or large print reading books.
Severe Low Vision	d. I can read with vision aids but slower than before. I use high power magnifiers.
Profound Low Vision	c. I may be able to read with vision aids but I prefer talking books.
Near-Blindness	b. I can see light, shadows, and movement. I cannot read using my vision. I rely on talking books, braille or other nonvisual sources.
Total Blindness	a. I do not see anything or I can only see light but no movement.

Supplementary Data S4 Method for calculating the salary gap between patients and the general population (yen/patient/year)

(i) The annual income of each patient who answered Question (Q) 22 of Module 2 (*n* = 37).

(ii) The employment rate of patients by sex and age group, which was calculated as the quotient of the number of patients who answered “Yes” to Q1 of Module 2 divided by the number of patients included in the analysis, stratified by sex and age group. The age groups were divided into 5-year brackets, per government data.

(iii) The result of multiplying each patient’s annual income and the employment rate of the corresponding sex and age group.

(iv) The average salary of the general population by sex and age group based on the Basic Survey on Wage Structure [1].

(v) The employment rate of the general population by sex and age group based on the Labour Force Survey [2].

(vi) The result of multiplying the average salary and the employment rate of the general population for each sex and age group.

(vii) The difference between the value calculated in (vi) of the patient’s sex and age group minus the value calculated in (iii) for each patient who answered Q22 of Module 2 (*n* = 37).

(viii) The quotient of the sum of the values calculated in (vii) divided by all patients included in the analysis (*N* = 122).

References

1. Ministry of Health, Labour and Welfare. Basic Survey on Wage Structure. https://www.mhlw.go.jp/english/database/db-l/wage-structure.html. Accessed 6 December 2022.

2. Statistics Bureau of Japan. Annual Report on the Labour Force Survey 2020. https://www.stat.go.jp/english/data/roudou/report/2020/index.html. Accessed 6 December 2022.

Supplementary Table S1 Retinitis pigmentosa patient out-of-pocket cost stratified by demographics

	n (%)	Out-of-pocket cost, mean (SD), yen/patient/year			
		Vision aids/modifications	Medications and supplements	Formal care	
				Healthcare services	Paid care
Overall	122 (100.0)	40,257 (103,470)	23,161 (113,260)	20,924 (68,717)	170,742 (1,252,264)
Age group, years					
≤ 64	86 (70.5)	43,088 (115,497)	26,302 (133,153)	26,309 (81,153)	183,312 (1,476,651)
> 64	34 (27.9)	34,582 (69,209)	11,283 (20,851)	7431 (9247)	148,990 (361,725)
Unknown	2 (1.6)	15,000 (21,213)	90,000 (127,279)	18,750 (7425)	0 (0)
Sex					
Female	67 (54.9)	56,015 (133,093)	29,619 (147,740)	23,524 (79,468)	245,042 (1,672,842)
Male	55 (45.1)	21,062 (40,696)	15,294 (44,173)	17,757 (53,326)	80,231 (279,989)
Employment					

Working	50 (41.0)	30,206 (48,661)	37,817 (173,222)	30,149 (102,234)	38,535 (120,538)
Student	1 (0.8)	2600 (-)	0 (-)	1740 (-)	0 (-)
Non-student	49 (40.2)	30,769 (49,001)	38,589 (174,930)	30,729 (103,210)	39,322 (121,658)
Non-working	72 (59.0)	47,238 (128,416)	12,983 (29,946)	14,518 (27,068)	262,553 (1,625,307)
Student	3 (2.5)	68,333 (114,054)	0 (0)	27,300 (25,159)	0 (0)
Non-student	69 (56.6)	46,320 (129,674)	13,548 (30,473)	13,962 (27,182)	273,968 (1,659,817)
Highest level of formal education					
Junior high school education	3 (2.5)	100,000 (91,652)	20,000 (34,641)	31,243 (17,952)	0 (0)
High school education	38 (31.1)	30,524 (59,393)	42,600 (194,336)	39,300 (117,598)	48,332 (122,904)
Bachelor’s degree	46 (37.7)	52,026 (148,440)	17,817 (51,062)	12,151 (22,690)	80,906 (296,970)
Master’s degree	7 (5.7)	21,715 (28,992)	9429 (9710)	10,057 (14,440)	5014 (13,267)
Doctoral degree	2 (1.6)	5500 (7778)	0 (0)	4855 (6413)	52,000 (73,539)
Others	26 (21.3)	34,435 (70,732)	10,048 (21,847)	12,560 (24,264)	582,045 (2,680,053)
Mode of inheritance					

Autosomal dominant	8 (6.6)	26,250 (35,832)	1200 (2566)	22,609 (39,244)	244,238 (682,963)
Autosomal recessive	17 (13.9)	34,706 (59,725)	13,835 (31,786)	58,994 (151,965)	18,353 (33,615)
X-linked	1 (0.8)	6000 (-)	0 (-)	30,000 (-)	40,430 (-)
Sporadic	57 (46.7)	37,228 (58,066)	28,815 (158,497)	17,815 (52,838)	310,618 (1,811,935)
Unknown	39 (32.0)	50,856 (164,828)	24,062 (56,528)	8295 (9217)	21,000 (66,144)
Physical disability grade					
Grade 1	45 (36.9)	53,800 (154,854)	33,938 (178,813)	7818 (10,789)	418,331 (2,049,064)
Grade 2	67 (54.9)	33,708 (57,535)	16,567 (44,743)	23,540 (79,615)	24,114 (86,823)
Grade 3	3 (2.5)	1000 (1732)	0 (0)	10,600 (7034)	130,000 (225,167)
Grade 4	0 (0.0)	-	-	-	-
Grade 5	5 (4.1)	29,800 (24,191)	19,200 (21,799)	111,448 (156,618)	0 (0)
Grade 6	0 (0.0)	-	-	-	-
No certificate	2 (1.6)	40,000 (56,569)	46,200 (19,516)	17,360 (13,350)	0 (0)
Distance vision impairment (WHO)					
Normal	25 (20.5)	32,552 (42,721)	60,240 (238,128)	63,773 (142,617)	3162 (15,808)

Mild	13 (10.7)	50,923 (72,387)	16,800 (50,027)	16,025 (15,099)	36,300 (107,720)
Moderate	10 (8.2)	4800 (8053)	18,720 (25,243)	21,064 (38,764)	0 (0)
Severe	4 (3.3)	36,250 (44,230)	6000 (6928)	4755 (4536)	34,125 (49,038)
Blindness	64 (52.5)	28,472 (51,006)	12,500 (41,860)	7435 (10,354)	311,283 (1,722,434)
Unknown	6 (4.9)	236,733 (390,459)	15,000 (14,071)	7432 (5370)	36,833 (52,270)
Visual impairment class (AMA)					
Near-normal vision	5 (4.1)	12,000 (21,679)	16,080 (17,298)	82,604 (167,322)	0 (0)
Moderate low vision	10 (8.2)	43,400 (60,206)	37,800 (93,640)	36,848 (34,533)	0 (0)
Severe low vision	21 (17.2)	11,895 (19,797)	8343 (15,877)	23,502 (40,631)	46,429 (149,843)
Profound low vision	23 (18.9)	43,217 (57,724)	18,313 (40,096)	35,350 (129,899)	12,593 (28,317)
Near-blindness	46 (37.7)	35,128 (64,481)	35,295 (176,696)	7842 (10,826)	389,738 (2,027,773)
Total blindness	17 (13.9)	91,629 (240,498)	8661 (23,428)	6113 (8065)	96,351 (195,312)

AMA American Medical Association, *SD* standard deviation, *WHO* World Health Organization

Supplementary Table S2 Robustness testing of the primary outcome, excluding patients with response inconsistencies

	Mean (SD)	Median	95% CI	Min, Max	Q1, Q3
Out-of-pocket cost, yen/patient/year[a]					
Vision aids/modification	31,222 (52,025)	10,000	[21,737, 40,707]	0, 300,000	0, 35,800
Medications and supplements (*n* = 117)	13,894 (36,556)	0	[7201, 20,588]	0, 300,000	0, 12,000
Formal care					
Healthcare services	13,514 (23,928)	5070	[9152, 17,877]	0, 151,580	1755, 14,475
Paid care (*n* = 117)	60,928 (210,205)	0	[22,437, 99,418]	0, 1,934,400	0, 26,000
Opportunity loss, yen/patient/year[a]					
Salary gaps with general population	650,705 (-)	-	-	-	-
Cost of depression/anxiety					
Direct medical costs	751 (-)	-	-	-	-
Suicide costs	12,161 (-)	-	-	-	-
Total annual cost, yen/patient[a]	783,176 (-)	-	-	-	-

Total lifetime cost[b], yen/patient[a]	66,389,827 (-)	-	-	-	-

[a] *N* = 118, unless otherwise specified

[b] Calculated by multiplying the total annual cost by 84.77 years (the average Japanese life expectancy in 2020)

CI confidence interval, *Q* quartile, *SD* standard deviation

Supplementary Table S3 Work Productivity and Activity Impairment Questionnaire: General Health V2.0 score stratified by demographics

	n (%)		WPAI:GH score, mean (SD)			
	a	b	Percent work time missed due to health[a] (Absenteeism)	Percent impairment while working due to health[a] (Presenteeism)	Percent overall work impairment due to health[a] (Work productivity loss)	Percent activity impairment due to health[b] (Activity impairment)
Overall	47 (100.0)	50 (100.0)	0.0 (0.0)	26.2 (33.2)	26.2 (33.2)	31.6 (35.0)
Age group						
18 to 64	43 (91.5)	45 (90.0)	0.0 (0.0)	26.0 (33.7)	26.0 (33.7)	32.9 (35.7)
> 64	4 (8.5)	5 (10.0)	0.0 (0.0)	27.5 (32.0)	27.5 (32.0)	20.0 (27.4)
Sex						
Female	20 (42.6)	22 (44.0)	0.0 (0.0)	23.0 (31.6)	23.0 (31.6)	35.9 (41.2)
Male	27 (57.4)	28 (56.0)	0.0 (0.0)	28.5 (34.7)	28.5 (34.7)	28.2 (29.4)

Mode of inheritance						
Autosomal dominant	1 (2.1)	1 (2.0)	0.0 (-)	0.0 (-)	0.0 (-)	10.0 (-)
Autosomal recessive	7 (14.9)	7 (14.0)	0.0 (0.0)	21.4 (31.3)	21.4 (31.3)	31.4 (35.8)
X-linked	0 (0.0)	0 (0.0)	—	—	—	—
Sporadic	23 (48.9)	25 (50.0)	0.0 (0.0)	25.2 (34.4)	25.2 (34.4)	29.6 (36.5)
Unknown	16 (34.0)	17 (34.0)	0.0 (0.0)	31.3 (34.2)	31.3 (34.2)	35.9 (34.8)
Employment						
Regular	21 (44.7)	22 (44.0)	0.0 (0.0)	39.0 (39.2)	39.0 (39.2)	42.7 (38.4)
Non-regular	16 (34.0)	17 (34.0)	0.0 (0.0)	8.8 (12.6)	8.8 (12.6)	24.7 (28.3)
Self-employed	10 (21.3)	11 (22.0)	0.0 (0.0)	27.0 (32.0)	27.0 (32.0)	20.0 (33.2)
Highest level of formal education						
High school education	14 (29.8)	14 (28.0)	0.0 (0.0)	28.6 (35.7)	28.6 (35.7)	32.1 (41.0)
Bachelor’s degree	19 (40.4)	19 (38.0)	0.0 (0.0)	23.7 (31.8)	23.7 (31.8)	37.9 (35.5)
Master’s degree	5 (10.6)	5 (10.0)	0.0 (0.0)	42.0 (49.2)	42.0 (49.2)	26.0 (36.5)

Others	9 (19.1)	12 (24.0)	0.0 (0.0)	18.9 (23.2)	18.9 (23.2)	23.3 (27.4)
Physical disability grade						
Grade 1	15 (31.9)	17 (34.0)	0.0 (0.0)	24.0 (33.6)	24.0 (33.6)	27.6 (36.5)
Grade 2	27 (57.4)	28 (56.0)	0.0 (0.0)	27.8 (34.3)	27.8 (34.3)	36.1 (35.3)
Grade 3	1 (2.1)	1 (2.0)	0.0 (-)	80.0 (-)	80.0 (-)	70.0 (-)
Grade 4	0 (0.0)	0 (0.0)	—	—	—	—
Grade 5	3 (6.4)	3 (6.0)	0.0 (0.0)	10.0 (10.0)	10.0 (10.0)	6.7 (11.5)
Grade 6	0 (0.0)	0 (0.0)	—	—	—	—
No certificate	1 (2.1)	1 (2.0)	0.0 (-)	10.0 (-)	10.0 (-)	10.0 (-)
Distance vision impairment (WHO)						
Normal	14 (29.8)	15 (30.0)	0.0 (0.0)	30.7 (33.2)	30.7 (33.2)	44.0 (36.6)
Mild	2 (4.3)	2 (4.0)	0.0 (0.0)	0.0 (0.0)	0.0 (0.0)	10.0 (14.1)
Moderate	3 (6.4)	3 (6.0)	0.0 (0.0)	33.3 (41.6)	33.3 (41.6)	36.7 (37.9)
Severe	1 (2.1)	1 (2.0)	0.0 (-)	0.0 (-)	0.0 (-)	0.0 (-)

Blindness	25 (53.2)	27 (54.0)	0.0 (0.0)	28.0 (34.8)	28.0 (34.8)	29.3 (34.9)
Unknown	2 (4.3)	2 (4.0)	0.0 (0.0)	0.0 (0.0)	0.0 (0.0)	0.0 (0.0)
Visual impairment class (AMA)						
Near-normal vision	3 (6.4)	3 (6.0)	0.0 (0.0)	6.7 (5.8)	6.7 (5.8)	3.3 (5.8)
Moderate low vision	5 (10.6)	5 (10.0)	0.0 (0.0)	22.0 (11.0)	22.0 (11.0)	32.0 (13.0)
Severe low vision	9 (19.1)	10 (20.0)	0.0 (0.0)	35.6 (35.0)	35.6 (35.0)	40.0 (32.0)
Profound low vision	9 (19.1)	9 (18.0)	0.0 (0.0)	43.3 (39.4)	43.3 (39.4)	47.8 (37.7)
Near-blindness	15 (31.9)	16 (32.0)	0.0 (0.0)	25.3 (37.4)	25.3 (37.4)	30.0 (38.6)
Total blindness	6 (12.8)	7 (14.0)	0.0 (0.0)	1.7 (4.1)	1.7 (4.1)	14.3 (37.8)

[a] Corresponds to patients with retinitis pigmentosa who are currently employed and have worked within the previous seven days

[b] Corresponds to patients with retinitis pigmentosa who are currently employed

AMA American Medical Association, *SD* standard deviation, *WHO* World Health Organization, *WPAI:GH* Work Productivity and Activity Impairment Questionnaire: General Health

Supplementary Table S4 Visual Function Questionnaire score stratified by visual impairment

	n (%)		NEI VFQ-25 score, mean (SD)												
			Composite	General health	General vision	Ocular pain	Near activities	Distance activities	Vision specific:				Driving	Color vision	Peripheral vision
	Except driving	Driving							Social functioning	Mental health	Role difficulties	Dependency			
Overall	118–122 (100.0)	63 (100.0)	42.0 (15.8)	51.0 (21.4)	38.7 (24.9)	78.9 (20.8)	31.5 (23.6)	38.6 (20.5)	43.7 (24.1)	36.6 (20.0)	48.7 (23.5)	41.6 (21.1)	6.3 (17.9)	57.0 (27.5)	23.5 (22.1)
Physical disability grade															
Grade 1	43–45 (36.1–36.9)	19 (30.2)	33.4 (11.4)	52.8 (23.4)	22.7 (23.6)	76.9 (22.1)	18.9 (15.4)	27.4 (15.6)	32.7 (20.2)	35.6 (20.8)	40.3 (23.8)	35.6 (17.0)	1.3 (5.7)	43.0 (27.5)	14.5 (19.1)
Grade 2	65–67 (54.9–55.5)	38 (60.3)	43.6 (13.3)	48.9 (19.7)	45.1 (19.8)	79.5 (20.6)	34.3 (22.1)	41.5 (18.8)	45.1 (21.4)	34.3 (18.0)	50.4 (20.8)	41.0 (20.1)	2.6 (11.7)	61.5 (23.4)	25.4 (21.7)

Grade 3	3 (2.5)	2 (3.2)	61.2 (11.2)	58.3 (14.4)	73.3 (11.5)	70.8 (7.2)	63.9 (12.7)	58.3 (8.3)	75.0 (12.5)	50.0 (6.3)	66.7 (26.0)	55.6 (4.8)	18.8 (26.5)	75.0 (25.0)	50.0 (0.0)
Grade 4	0 (0.0)	0 (0.0)	—	—	—	—	—	—	—	—	—	—	—	—	—
Grade 5	5 (4.1–4.2)	3 (4.8)	72.8 (12.6)	55.0 (20.9)	64.0 (16.7)	90.0 (16.3)	71.7 (18.3)	70.0 (12.6)	87.5 (12.5)	61.3 (25.5)	77.5 (18.5)	78.3 (20.1)	54.2 (19.1)	90.0 (13.7)	50.0 (17.7)
Grade 6	0 (0.0)	0 (0.0)	—	—	—	—	—	—	—	—	—	—	—	—	—
No certificate	2 (1.6–1.7)	1 (1.6)	76.7 (9.5)	62.5 (53.0)	70.0 (14.1)	87.5 (17.7)	75.0 (23.6)	83.3 (0.0)	81.3 (8.8)	53.1 (30.9)	81.3 (8.8)	83.3 (11.8)	75.0 (-)	100.0 (0.0)	50.0 (0.0)
Polyserial correlation coefficient[a]			0.628[b]	−0.036	0.603[b]	0.101	0.606[b]	0.577[b]	0.546[b]	0.175	0.401[b]	0.395	0.561[b]	0.504[b]	0.443[b]
Distance vision impairment (WHO)															
Normal	25	13	58.8	52.0	58.4	79.5	59.8	59.5	66.5	44.0	64.5	56.7	21.2	81.0	36.0

	(20.5–21.2)	(20.6)	(15.7)	(20.3)	(19.9)	(18.7)	(21.3)	(16.9)	(20.3)	(22.9)	(23.0)	(21.5)	(29.9)	(18.1)	(19.2)
Mild	13 (10.7–11.0)	7 (11.1)	44.5 (10.3)	44.2 (20.8)	53.8 (17.1)	76.9 (20.3)	41.7 (21.8)	46.8 (15.8)	50.0 (18.4)	31.3 (15.3)	43.3 (18.1)	37.8 (18.5)	0.0 (0.0)	61.5 (16.5)	23.1 (16.0)
Moderate	10 (8.2–8.5)	7 (11.1)	46.0 (13.6)	45.0 (19.7)	48.0 (16.9)	81.3 (18.9)	25.8 (14.4)	43.3 (20.7)	55.0 (20.6)	38.1 (21.5)	45.0 (25.1)	49.2 (28.5)	0.0 (0.0)	72.5 (18.4)	35.0 (29.3)
Severe	4 (3.3–3.4)	3 (4.8)	25.5 (5.7)	43.8 (23.9)	20.0 (0.0)	78.1 (29.5)	12.5 (10.8)	19.8 (6.3)	18.8 (16.1)	17.2 (10.7)	37.5 (28.9)	25.0 (18.0)	0.0 (0.0)	37.5 (25.0)	6.3 (12.5)
Blindness	61–64 (51.3–52.5)	29 (46.0)	36.9 (11.8)	55.1 (20.5)	29.4 (23.6)	81.3 (19.5)	22.1 (16.5)	31.2 (16.1)	35.3 (19.9)	36.9 (18.9)	45.9 (22.5)	37.8 (16.7)	3.4 (13.3)	45.9 (26.3)	18.0 (20.5)
Unknown	5–6	4 (6.3)	25.8 (13.1)	33.3 (30.3)	20.0 (17.9)	52.1 (29.0)	13.9 (12.5)	16.7 (14.9)	20.8 (18.8)	24.0 (20.3)	37.5 (17.7)	26.4 (23.8)	6.3 (12.5)	45.0 (32.6)	20.8 (29.2)

	(4.2–5.0)														
Polyserial correlation coefficient[a]			−0.599[b]	0.134	−0.589[b]	0.058	−0.667[b]	−0.613[b]	−0.576[b]	−0.111	−0.299	−0.360	−0.375	−0.614[b]	−0.361
Visual impairment class (AMA)															
Near-normal vision	5 (4.1–4.2)	2 (3.2)	75.1 (11.5)	65.0 (22.4)	68.0 (17.9)	90.0 (10.5)	83.3 (11.8)	75.0 (11.8)	82.5 (11.2)	61.3 (16.2)	85.0 (10.5)	78.3 (22.5)	37.5 (53.0)	95.0 (11.2)	45.0 (11.2)
Moderate low vision	10 (8.2–8.5)	7 (11.1)	61.4 (9.9)	47.5 (18.4)	58.0 (17.5)	85.0 (17.5)	55.8 (13.1)	61.3 (12.4)	76.3 (10.9)	53.1 (20.9)	60.0 (15.4)	63.3 (24.3)	17.9 (22.7)	82.5 (12.1)	52.5 (21.9)
Severe low vision	21 (17.2–17.8)	12 (19.0)	50.0 (12.7)	48.8 (18.5)	57.1 (20.3)	74.4 (18.7)	51.4 (19.0)	51.2 (14.7)	53.6 (19.8)	34.8 (17.7)	54.2 (24.2)	45.6 (15.5)	3.1 (10.8)	72.6 (19.2)	28.6 (18.2)
Profound low vision	22–23	15 (23.8)	40.5 (11.1)	48.9 (17.6)	46.1 (12.7)	78.3 (18.5)	25.0 (16.3)	38.8 (17.0)	47.3 (16.4)	27.4 (16.6)	44.6 (19.9)	37.7 (18.8)	5.0 (19.4)	58.0 (21.0)	25.0 (16.9)

	(18.6–19.3)														
Near-blindness	43–46 (36.4–38.0)	21 (33.3)	33.6 (13.0)	48.4 (24.4)	27.4 (19.5)	76.6 (24.8)	19.9 (16.1)	26.4 (16.8)	28.8 (20.4)	33.8 (20.1)	41.0 (23.2)	33.3 (18.8)	3.0 (13.6)	44.8 (25.3)	17.0 (21.4)
Total blindness	16–17 (13.2–14.4)	6 (9.5)	35.8 (7.1)	61.8 (20.0)	16.5 (26.7)	84.6 (16.8)	17.6 (12.8)	31.4 (13.7)	35.9 (16.4)	41.5 (16.8)	50.7 (22.3)	40.7 (12.5)	4.2 (10.2)	41.2 (29.2)	7.8 (15.1)
Polyserial correlation coefficient[a]			−0.641[b]	0.079	−0.650[b]	−0.008	−0.702[b]	−0.615[b]	−0.612[b]	−0.153	−0.284	−0.413[b]	−0.288	−0.581[b]	−0.525[b]

[a] Polyserial correlation coefficient between the level of visual impairment and the VFQ-25 score

[b] Correlation observed between the level of visual impairment and the VFQ-25 score

AMA American Medical Association, *NEI VFQ-25* National Eye Institute Visual Function Questionnaire-25, *SD* standard deviation, *WHO* World Health Organization

Supplementary Table S5 Health Utilities Index Mark 3 score stratified by visual impairment

	n (%)	HUI3 score, mean (SD)								
		Overall utility	Vision	Hearing	Speech	Ambulation	Dexterity	Emotion	Cognition	Pain
Overall	122 (100.0)	0.393 (0.247)	0.350 (0.362)	0.970 (0.125)	0.940 (0.154)	0.814 (0.221)	0.976 (0.127)	0.876 (0.175)	0.893 (0.186)	0.898 (0.167)
Physical disability grade										
Grade 1	45 (36.9)	0.255 (0.169)	0.059 (0.139)	0.966 (0.096)	0.959 (0.102)	0.734 (0.269)	0.960 (0.189)	0.885 (0.170)	0.878 (0.203)	0.898 (0.206)
Grade 2	67 (54.9)	0.427 (0.216)	0.455 (0.321)	0.982 (0.090)	0.918 (0.188)	0.842 (0.178)	0.983 (0.073)	0.859 (0.186)	0.903 (0.182)	0.892 (0.148)
Grade 3	3 (2.5)	0.827 (0.229)	0.967 (0.029)	1.000 (0.000)	1.000 (0.000)	0.943 (0.098)	1.000 (0.000)	1.000 (0.000)	0.900 (0.173)	0.897 (0.117)
Grade 4	0 (0.0)	-	-	-	-	-	-	-	-	-

Grade 5	5	0.780	0.950	0.800	1.000	1.000	1.000	0.964	0.924	0.952
	(4.1)	(0.300)	(0.000)	(0.447)	(0.000)	(0.000)	(0.000)	(0.049)	(0.130)	(0.044)
Grade 6	0	-	-	-	-	-	-	-	-	-
	(0.0)									
No certificate	2	0.756	0.950	1.000	1.000	1.000	1.000	0.865	0.850	0.960
	(1.6)	(0.307)	(0.000)	(0.000)	(0.000)	(0.000)	(0.000)	(0.191)	(0.212)	(0.057)
Polyserial correlation coefficient[a]		0.622[b]	0.834[b]	−0.097	−0.042	0.385	0.135	0.041	0.082	0.028
Distance vision impairment (WHO)										
Normal	25	0.628	0.792	0.979	0.932	0.900	0.968	0.880	0.915	0.912
	(20.5)	(0.267)	(0.251)	(0.104)	(0.174)	(0.143)	(0.113)	(0.143)	(0.166)	(0.071)
Mild	13	0.397	0.652	0.923	0.935	0.898	1.000	0.795	0.772	0.805
	(10.7)	(0.308)	(0.250)	(0.277)	(0.128)	(0.144)	(0.000)	(0.295)	(0.302)	(0.278)
Moderate	10	0.519	0.471	1.000	0.949	0.867	0.988	0.919	0.946	0.929
	(8.2)	(0.125)	(0.151)	(0.000)	(0.113)	(0.152)	(0.038)	(0.028)	(0.094)	(0.067)

Severe	4	0.384	0.285	1.000	1.000	0.715	1.000	0.865	0.965	0.940
	(3.3)	(0.107)	(0.190)	(0.000)	(0.000)	(0.272)	(0.000)	(0.090)	(0.070)	(0.040)
Blindness	64	0.297	0.119	0.970	0.935	0.773	0.981	0.892	0.905	0.918
	(52.5)	(0.164)	(0.221)	(0.101)	(0.168)	(0.246)	(0.126)	(0.165)	(0.164)	(0.144)
Unknown	6	0.230	0.162	0.953	0.970	0.693	0.867	0.813	0.810	0.742
	(4.9)	(0.279)	(0.259)	(0.072)	(0.073)	(0.289)	(0.327)	(0.237)	(0.264)	(0.368)
Polyserial correlation coefficient[a]		−0.578[b]	−0.806[b]	−0.003	0.001	−0.351	0.027	0.072	0.032	0.078
Visual impairment class (AMA)										
Near-normal vision	5	0.918	0.960	1.000	1.000	1.000	1.000	0.964	0.984	0.968
	(4.1)	(0.075)	(0.022)	(0.000)	(0.000)	(0.000)	(0.000)	(0.049)	(0.036)	(0.044)
Moderate low vision	10	0.564	0.756	0.900	0.908	1.000	0.933	0.837	0.924	0.885
	(8.2)	(0.293)	(0.236)	(0.316)	(0.203)	(0.000)	(0.174)	(0.296)	(0.122)	(0.157)
Severe low vision	21	0.484	0.658	0.975	0.851	0.841	0.994	0.887	0.859	0.896
	(17.2)	(0.236)	(0.293)	(0.113)	(0.256)	(0.133)	(0.026)	(0.155)	(0.212)	(0.077)

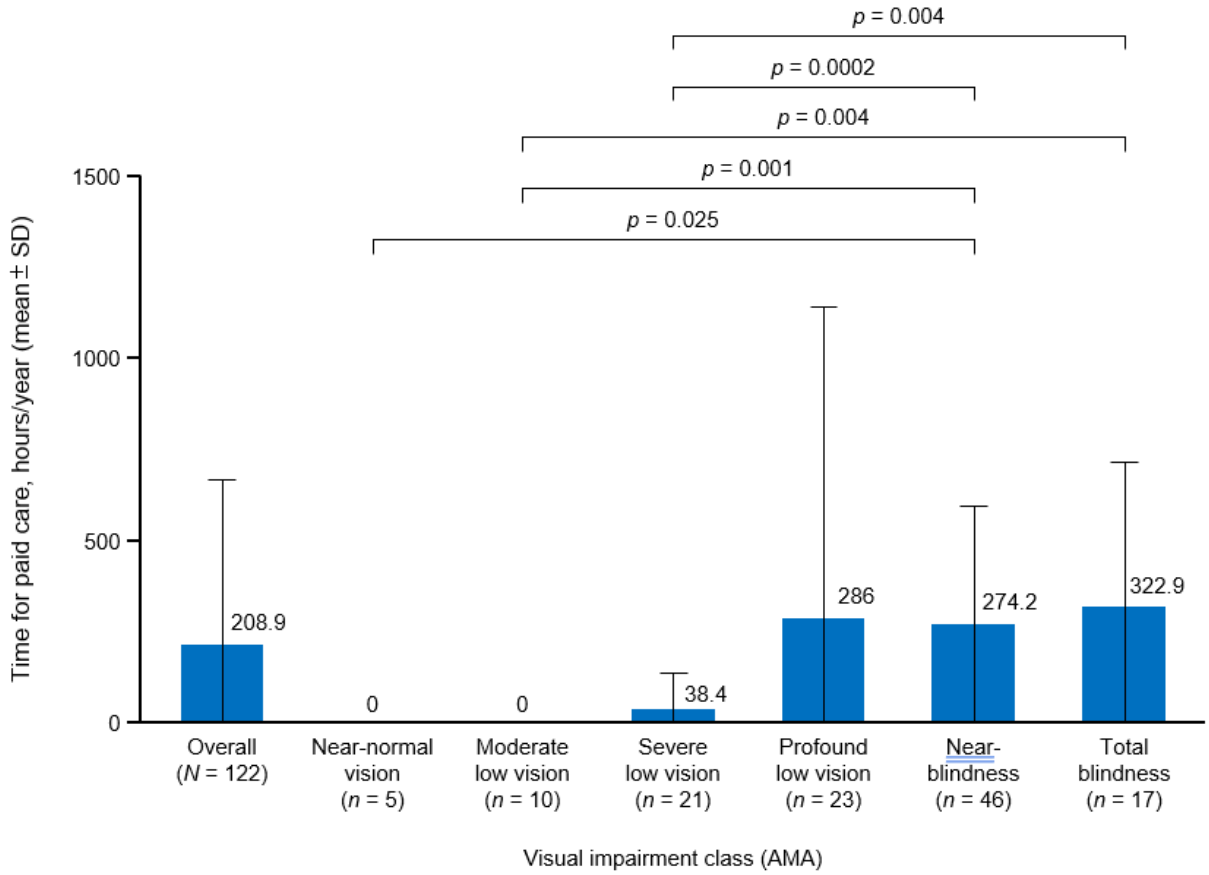
Profound low vision	23	0.378	0.399	1.000	0.971	0.812	0.984	0.820	0.861	0.851
	(18.9)	(0.242)	(0.275)	(0.000)	(0.095)	(0.203)	(0.041)	(0.253)	(0.247)	(0.273)
Near-blindness	46	0.318	0.160	0.962	0.957	0.778	0.961	0.883	0.912	0.900
	(37.7)	(0.184)	(0.244)	(0.117)	(0.117)	(0.260)	(0.187)	(0.131)	(0.140)	(0.165)
Total blindness	17	0.252	0.000	0.975	0.959	0.718	1.000	0.921	0.885	0.944
	(13.9)	(0.118)	(0.000)	(0.055)	(0.095)	(0.233)	(0.000)	(0.063)	(0.227)	(0.060)
Polyserial correlation coefficient[a]		−0.536[b]	−0.769[b]	0.025	0.115	−0.351	0.021	0.056	−0.020	0.056

[a] Polyserial correlation coefficient between the level of visual impairment and the HUI3 score

[b] Correlation observed between the level of visual impairment and the HUI3 score

AMA American Medical Association, *HUI3* Health Utilities Index Mark 3, *SD* standard deviation, *WHO* World Health Organization

Supplementary Fig. S1 Time for paid care received by patients with retinitis pigmentosa, stratified by vision impairment class



Statistical significance was determined according to the Steel–Dwass test.

AMA American Medical Association, *SD* standard deviation