Economic value of early genetic testing in inherited retinal dystrophy diagnosis

Qiaoyi Zhang,[1] Qian Cai,[1] Kirsten Lum,[2] Wei Wang,[2] Ting Wu,[2] Pawel Skiba,[1] Juhyeon Song,[1,*] Marvin Sperling[1]

[1]Janssen Global Services, LLC, Raritan, NJ, USA; [2]Data Sciences, Janssen Global Services, LLC, Titusville, NJ, USA.

*Presenting author.

BACKGROUND

- Inherited retinal dystrophy (IRD), which is characterized by photoreceptor cell death and loss of the retinal pigment epithelium, is a diverse group of diseases that can cause severe vision loss or blindness
- Genetic testing is a critical step for obtaining a confirmatory diagnosis for patients with IRD and enabling access to eligible treatment and care
- A lack of awareness and reimbursement for genetic testing prevents many patients from receiving timely genetic testing, which results in delayed diagnosis or misdiagnosis of IRD

OBJECTIVE

- To assess healthcare resource utilization (HCRU) and costs associated with the IRD diagnostic journey based on the timing of genetic testing (early vs delayed testing)

METHODS

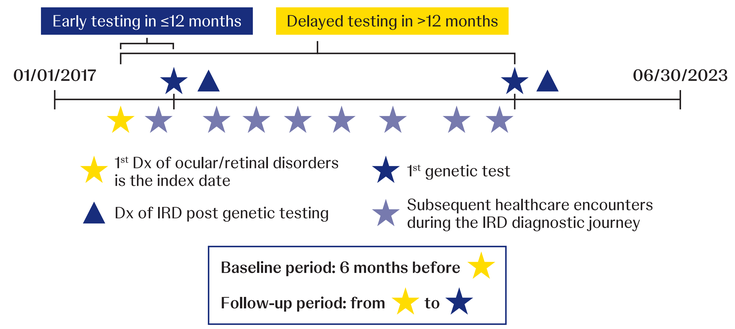
Data Source

- This retrospective cohort study used Optum's de-identified Clinformatics® Data Mart database to select eligible patients in the United States; this database contains the socioeconomic status and HCRU for individuals with both medical and pharmacy coverage at the US Census Division level

Study Cohort

- Patients were categorized into the early testing group if their first genetic test was within 12 months of the index date; otherwise, they were categorized into the delayed testing group (**Figure 1**)
- All-cause and ocular/retinal disorder–related HCRU and costs incurred for medical and pharmacy services from the index date to the first genetic test date were assessed

Figure 1. Study design.

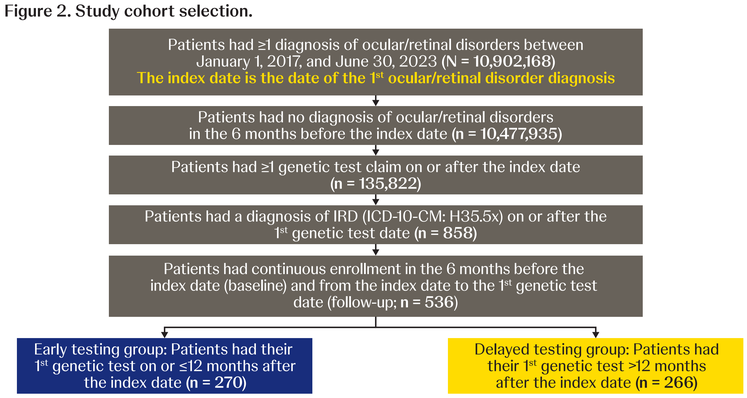


Dx, diagnosis, IRD, inherited retinal dystrophy.

Statistical Analyses

- Descriptive analysis was conducted using R statistical software (version 4.3.1). Differences in continuous and categorical variables between the 2 groups were compared using *t* tests, Fisher's exact tests, or chi-square tests, as appropriate

Figure 2. Study cohort selection.



ICD-10-CM, *International Classification of Diseases, Tenth Revision, Clinical Modification*; IRD, inherited retinal dystrophy.

RESULTS

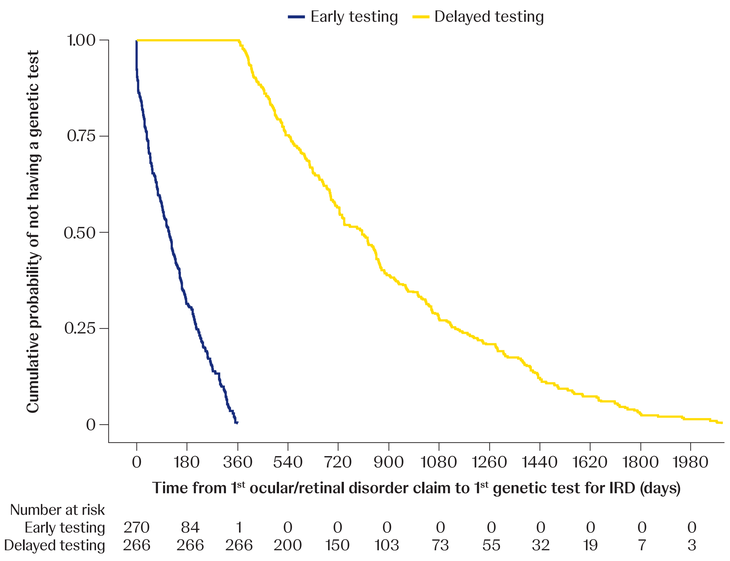
- A total of 536 patients were included (**Figure 2**). The mean ± standard deviation (SD) age was 55 ± 21.2 years, the median age was 62 years, and 64% of patients were female. The patients in the delayed testing group were older than those in the early testing group (P <0.001; **Table 1**)
- Median time from the first ocular/retinal disorder diagnosis to the first genetic test was 116 days for the early testing group and 805 days for the delayed testing group (**Figure 3**)
- During the diagnostic journey, patients in the early testing group incurred mean ± SD total healthcare costs of $13,084 ± $30,912 and mean ocular disorder–related costs of $2689 ± $7811. Patients in the delayed testing group had 5 times higher mean all-cause costs of $76,838 ± $116,372 and almost 3 times higher mean ocular disorder–related costs of $7830 ± $25,050 compared to patients in the early testing group
- Compared to patients in the early testing group, patients in the delayed testing group had 3 times more physician visits for ocular/retinal disorders and had more visits with ophthalmologists, optometrists, other specialists, and primary care providers for ocular/retinal disorders during the diagnostic journey (**Figure 4**)
- The average cost paid by insurance for observed IRD genetic testing ranged from $203 to $1559 (**Table 2**)

Table 1. Demographic Characteristics of Study Patients

	Early testing (n = 270)	Delayed testing (n = 266)
Age at index date,* years, mean ± SD	52 ± 21.7	59 ± 20.2
Female, %	60	67
Race/ethnicity, %		
African American	12	11
Asian	4	2
White	73	76
Hispanic	7	7
Other/unknown	5	5
Region,* %		
Northeast	16	20
Midwest	26	16
South	46	47
West	13	17

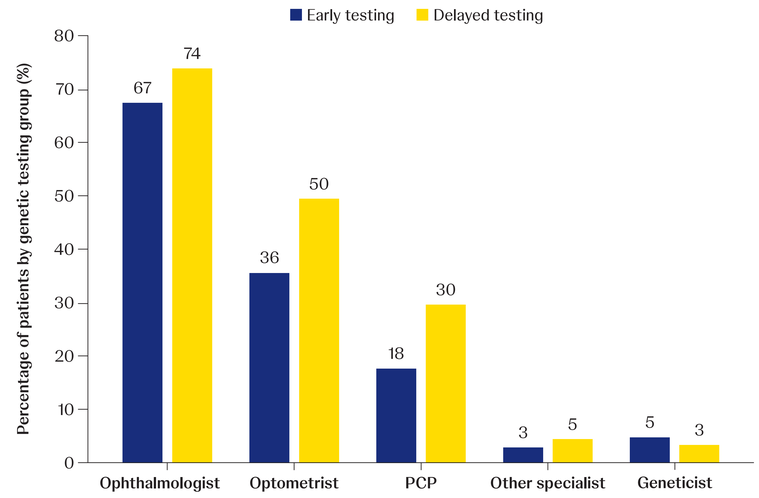
SD, standard deviation.
*The difference between the early and delayed testing groups was statistically significant (P <0.05).

Figure 3. Time from the first ocular/retinal disorder claim to the first genetic test for IRD.



IRD, inherited retinal dystrophy.

Figure 4. Distribution of the type of healthcare provider visited for ocular/retinal disorders during the IRD diagnostic journey.



IRD, inherited retinal dystrophy; PCP, primary care provider.

Table 2. Average Cost Paid by Insurance for the First Genetic Test of the IRD Genetic Diagnosis

Genetic test procedure code	Procedure description	Average cost paid by insurance ($USD)
81400	Molecular pathology procedure, level 1	203.16
81401	Molecular pathology procedure, level 2	302.13
81404	Molecular pathology procedure, level 5	457.14
81405	Molecular pathology procedure, level 6	488.08
81406	Molecular pathology procedure, level 7	443.20
81407	Molecular pathology procedure, level 8	1085.53
81408	Molecular pathology procedure, level 9	1559.33
81434	Hereditary retinal disorders	495.11
81479	Unlisted molecular pathology procedure	629.52
81599	Unlisted multianalyte assay with algorithmic analysis	961.50

IRD, inherited retinal dystrophy; USD, United States dollar.

CONCLUSIONS

- Descriptive data showed that patients with delayed genetic testing had substantially higher all-cause and ocular disorder–related healthcare costs than those who received early genetic testing during their IRD diagnostic journey
- The findings from this study suggest that early genetic testing in patients with IRD shortens the diagnostic journey and may reduce the associated healthcare costs

Disclosures

All authors are employees of Janssen Global Services, LLC.

Acknowledgments

This study was sponsored by Janssen Global Services, LLC. Technical editorial support was provided by Alanna Kahhan, ELS, of Lumanity Communications Inc., and was funded by Janssen Global Services, LLC.

An electronic version of the poster can be viewed by scanning the QR code. The QR code is intended to provide scientific information for individual reference. The PDF should not be altered or reproduced in any way.

Poster presented at the Professional Society for Health Economics and Outcomes Research (ISPOR) Conference; May 5-8, 2024; Atlanta, GA, USA.