Difficulty in performing everyday activities in patients with juvenile macular dystrophies: comparison with patients with retinitis pigmentosa

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Abstract

***Aims*—To ascertain the level of perceived difficulty experienced by patients with central vision loss due to juvenile macular dystrophies in the performance of everyday activities. A second objective was to compare their perceived difficulty with that of patients with retinitis pigmentosa (RP) with primarily peripheral vision loss.**

***Methods*—72 patients with Stargardt disease, cone dystrophy, or cone-rod dystrophy who had visual acuities worse than 20/40 and normal peripheral visual fields rated themselves on their difficulty in the performance of 33 activities encompassing a wide variety of everyday tasks. These findings were compared with the responses of 120 patients with typical RP or Usher syndrome type 2 who had visual acuities of 20/40 or better and peripheral visual field loss.**

***Results*—The juvenile macular dystrophy group reported the greatest level of overall self perceived difficulty with activities involving central vision, and lesser and variable degrees of difficulty with items within the mobility, negotiating steps, driving, and miscellaneous categories. Consistent with these findings, there were highly significant correlations between subjects' rated performances of activities involving central vision and the clinical measures of vision, including visual acuity and size of central scotoma. There were fewer significant correlations between perceived performance of activities in the other categories and the clinical measures. In general, those activities that showed significant correlations with the clinical measures of vision for the patients with juvenile macular dystrophies also showed significant differences in the patterns of responses between the juvenile macular dystrophy group and the RP group. Those items which were not correlated with the clinical measures in the juvenile macular dystrophy group tended not to show significant differences in the response patterns between the two groups.**

***Conclusion*—These results provide insight into the types of perceived difficulties in performing tasks of everyday life in patients with these disorders which affect counselling of these patients.**

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The loss of visual function in retinal disorders is typically assessed through clinical tests such as visual acuity, visual fields, and electroretinography. The impact of the loss of visual function on the performance of everyday tasks and the relation of visual function to these clinical tests is not well understood. One approach has been to administer questionnaires regarding self perceived difficulties. These questionnaires have been shown to correlate with actual difficulty in performance.[1]

In a study of patients with retinitis pigmentosa (RP), we found that patients had a wide range of perceived difficulty in everyday tasks, and the perceived difficulty correlated significantly with visual acuity, less so with visual fields and electroretinogram amplitudes.[2] This finding is consistent with the results from studies of aging which have found that visual acuity is a risk factor predictive of perceived difficulty among older populations.[3 4]

Given that visual acuity seems to be related to self perceived difficulty with everyday tasks, it seems logical that patients who have central vision loss would report considerable difficulty with these tasks. It is less clear how much difficulty would be reported with tasks that involve peripheral vision. Further, intuitively, patients afflicted with a disease such as RP, in which peripheral field loss can be severe in the presence of good functional central vision, are likely to be more encumbered by performing tasks necessitating good peripheral field function, compared with patients with macular disease and reduced central visual function but normal peripheral fields. To investigate this issue, we examined the self reported difficulty of patients with one of three juvenile onset macular dystrophies—Stargardt disease, cone dystrophy, or cone-rod dystrophy; with visual acuities worse than 20/40, and intact peripheral visual fields. These results were then compared with the results from a group of RP patients, a subset of those reported previously,[2] with visual acuities of 20/40 or better and peripheral visual field loss. Patients with these juvenile macular dystrophies often have central visual field scotomas, reduced visual acuity, and colour vision abnormalities.[5–7] RP is characterised by a progressive loss of visual field, typically in the mid-periphery and far periphery; pigmentary changes of the retina; and poor vision in dim light.[8]

Patients and methods

PATIENTS

Seventy two patients with central vision loss (36 women, 36 men) due to Stargardt disease

*Table 1 Responses of patients with juvenile macular dystrophies correlated with clinical vision measures**

Activity	*VA*	*Scot II-4-e*
Activities involving central vision:		
Threading a needle	0.47***	−0.38***
Reading street signs at night	0.46***	−0.42***
Reading directions on medicine bottles	0.35***	−0.20**
Reading ingredients on cans of food	0.30***	−0.18
Reading street signs during the day	0.49***	−0.47***
Seeing faces across a street	0.46***	−0.29***
Reading ordinary newspaper print	0.64***	−0.52***
Playing cards	0.52***	−0.47***
Reading numbers on TV screen	0.29***	−0.32***
Finding destination/unfamiliar buildings	0.13	−0.15
Mobility:		
Finding a seat in a movie theatre	0.25**	−0.34***
Finding particular items in a store	0.24**	−0.16
Walking outdoors at night	0.30***	−0.36***
Participating in social gatherings	0.11	−0.09
Walking through shopping malls	0.16	−0.21**
Negotiating steps:		
Walking down steps in dim light	0.27**	−0.27**
Walking down steps during daylight	0.26**	−0.40***
Driving:		
Driving at night	0.12	0.19
Driving during the day	−0.20	−0.10
Driving in unfamiliar areas	0.40**	0.02
Eating:		
Eating meals	0.07	−0.10
Miscellaneous:		
Writing checks	0.51***	−0.41***
Engaging in physical exercise	0.22**	−0.19
Watching a movie at a theatre	0.23**	−0.35***
Using a ruler	0.36***	−0.32***
Walking outdoors during the day	0.05	−0.20**
Preparing meals	0.04	−0.06
Grooming	−0.11	−0.06
Using a screwdriver	0.07	0.10
Using escalators	−0.04	−0.08
Watching television	0.09	−0.37***
Grocery shopping	0.24**	−0.28***
Using public transport	0.29**	−0.26**

*VA indicates visual acuity; Scot II-4-e, areas of central scotoma to the II-4-e Goldmann targets.
**$0.01<p\leq0.05$.
***$p\leq0.01$.

(n = 47), cone dystrophy (n = 7), or cone-rod dystrophy (n = 18) participated in this study. They ranged in age from 12 to 74 years with a mean age of 37.3 years (SD 14.2). Their Snellen visual acuities ranged from 20/50 to 20/800 in the better eye, with a median visual acuity of 20/200. All patients showed normal peripheral field boundaries to the II-4-e target of the Goldmann perimeter.

Also included were the visual acuity, visual field, and questionnaire data from a subset of 120 patients with RP (54 women and 66 men) from a previous study.[2] The patients with RP ranged in age from 12 to 76 years, with a mean age of 39.9 years (SD 13.7 years), were selected on the basis of visual acuity, and had minimal or no cataracts. Their Snellen visual acuities ranged from 20/15 to 20/40 in their better eye, with a median of 20/20. The distribution of genetic types for the 120 patients with RP was as follows: autosomal dominant (n = 21, 17.5%); autosomal recessive (n = 12, 10.0%); X linked recessive (n = 6, 5.0%); isolated (n = 59, 49.2%); indeterminate (n = 15, 12.5%); and Usher syndrome type 2 (n = 7, 5.8%).

These groups of patients resulted from screening the files of all patients with these diagnoses within a referral clinic of a state university medical centre. Those patients who were available for testing and fulfilled our vision criteria participated. The patients represent a wide variety of racial and socioeconomic levels given that the clinic patients are sampled primarily from urban and suburban environments throughout Illinois and surrounding regions (for example, Wisconsin, Indiana, and Michigan).

For patients in both groups, visual fields were obtained for each eye using a Goldmann perimeter with the II-4-e target for the patients with juvenile macular dystrophies and for the patients with RP. We calculated visual field areas using computerised planimetry as described previously.[9] For the patients with juvenile macular dystrophies, the mean value for the area of central scotoma in the better eye was 677.4 mm^2 (SD 100.64 mm^2) for the II-4-e target. The mean visual field area for the patients with RP was 2929.0 mm^2 (398.06 mm^2) to the II-4-e target. The mean visual field area for the II-4-e target for a group of 21 normally sighted individuals was 17 758.3 mm^2 (1671.0 mm^2). For comparison, the areas of visual field diameters of 20° and 40° are 473.4 mm^2 and 1753.0 mm^2, respectively.

METHODS

Self report questionnaire

To assess patients' self perceived functioning on selected everyday activities, we used a 33 item questionnaire described in detail in a previous study.[2] The questionnaire was administered to the patients verbally by one of the authors (BIR). The format of the questions was based on that of the ADVS questionnaire developed by Mangione *et al.*[10] Subjects were asked to rate themselves as to the level of difficulty they experienced in performing each activity (none, mild, moderate, extreme, and not able) within the past 2 years. If patients responded that they did not perform an activity for reasons other than visual ones, they were excluded from the analysis for that question. The activities are listed in Table 1, and the items are categorised according to the factor analysis from our previous study.[2] The questionnaire was administered to a group of 10 normally sighted individuals. Because all the normal subjects reported having no difficulty on all of the items, we reasoned that it would not be productive to pursue any analyses comparing the patients with the normally sighted group.

Results

We performed a factor analysis of the responses to the questionnaire items for the patients with juvenile macular dystrophies. The six factors that were identified by the factor analysis were similar to those in our previous study of patients with RP. Because of this similarity, and for ease of comparison, we present our results in the categories from the previous study.

We compared patients' responses with their clinical vision measures. In Table 1, we include the Spearman correlation coefficients between the juvenile macular dystrophy patients' questionnaire responses and the visual acuities of these patients' better eyes, and the patients' responses and the areas of central scotoma to the II-4-e test target in the patients' better eyes. There was a statistically significant correlation

Table 2 Percentage of subjects with juvenile macular dystrophy or retinitis pigmentosa responding to activities at each level of difficulty

Activity	χ^2	df	p Value	Level of difficulty: None	Mild	Moderate	Extreme	Not able	Did not perform for non-visual reasons	Group*
Activities involving central vision:										
Threading a needle	32.8	4	0.001***	2.8	12.5	1.4	13.9	45.8	23.6	JMD
				20.0	15.0	13.3	5.0	18.3	28.3	RP
Reading street signs at night	57.7	4	0.001***	0.0	9.7	15.3	12.5	62.5	0.0	JMD
				14.2	25.0	27.5	20.0	11.7	1.7	RP
Reading directions on medicine bottles	59.9	4	0.001***	8.3	12.5	15.3	19.4	44.4	0.0	JMD
				39.2	28.3	13.3	11.7	4.2	3.3	RP
Reading ingredients on cans of food	88.4	4	0.001***	6.9	8.3	15.3	19.4	45.8	4.2	JMD
				51.7	25.0	12.5	4.2	2.5	4.2	RP
Reading street signs during the day	79.1	4	0.001***	2.8	11.1	26.4	20.8	38.9	0.0	JMD
				39.2	33.3	19.2	5.8	2.5	0.0	RP
Seeing faces across a street	67.6	4	0.001***	2.8	11.1	5.6	0.0	79.2	1.4	JMD
				40.0	25.8	10.0	2.5	20.8	0.8	RP
Reading ordinary newspaper print	84.9	4	0.001***	5.6	19.4	15.3	8.3	50.0	1.4	JMD
				55.0	29.2	10.0	1.7	3.3	0.8	RP
Playing cards	49.0	4	0.001***	19.4	33.3	12.5	5.6	16.7	12.5	JMD
				59.2	11.7	5.0	5.8	0.0	18.3	RP
Reading numbers on TV screen	56.8	4	0.001***	19.4	29.2	20.8	5.6	23.6	1.4	JMD
				70.8	20.0	5.8	0.8	2.5	0.0	RP
Finding destination/unfamiliar buildings	21.8	3	0.001***	15.3	37.5	29.2	16.7	0.0	1.4	JMD
				46.7	31.7	14.2	7.5	0.0	0.0	RP
Mobility:										
Finding a seat in a movie theatre	35.5	3	0.001***	45.8	18.1	8.3	13.9	0.0	13.9	JMD
				11.7	12.5	28.3	30.0	0.0	17.5	RP
Finding particular items in a store	8.7	3	0.034**	31.9	29.2	18.1	5.6	0.0	15.3	JMD
				51.7	25.0	7.5	5.0	0.0	10.8	RP
Walking outdoors at night	35.9	4	0.001***	52.8	18.1	11.1	2.8	13.9	1.4	JMD
				14.2	24.2	25.8	15.0	18.3	2.5	RP
Participating in social gatherings	4.0	4	0.408	51.4	29.2	13.9	0.0	2.8	2.8	JMD
				41.7	33.3	19.2	2.5	1.7	1.7	RP
Walking through shopping malls	27.9	4	0.001***	70.8	13.9	11.1	2.8	1.4	0.0	JMD
				31.7	29.2	22.5	9.2	6.7	0.8	RP
Negotiating steps:										
Walking down steps in dim light	42.6	4	0.001***	45.8	29.2	2.8	0.0	20.8	1.4	JMD
				11.7	17.5	22.5	6.7	34.2	7.5	RP
Walking down steps during daylight	18.4	4	0.001***	62.5	22.2	2.8	0.0	11.1	1.4	JMD
				34.2	24.2	12.5	5.8	18.3	5.0	RP
Driving:										
Driving at night	5.3	4	0.259	5.6	9.7	5.6	1.4	13.9	63.9	JMD
				11.7	18.3	16.7	13.3	16.7	23.3	RP
Driving during the day	15.0	4	0.005***	5.6	23.6	2.8	2.8	1.4	63.9	JMD
				37.5	29.2	10.0	1.7	0.0	21.7	RP
Driving in unfamiliar areas	10.2	4	0.038**	0.0	11.1	11.1	2.8	8.3	66.7	JMD
				15.0	30.8	12.5	6.7	8.3	26.7	RP
Eating:										
Eating meals	1.8	2	0.420	88.9	9.7	1.4	0.0	0.0	0.0	JMD
				86.7	8.3	5.0	0.0	0.0	0.0	RP
Miscellaneous:										
Writing checks	36.4	4	0.001***	40.3	16.7	5.6	6.9	12.5	18.1	JMD
				83.3	5.0	1.7	0.8	1.7	7.5	RP
Engaging in physical exercise	2.8	4	0.587	65.3	19.4	4.2	0.0	4.2	6.9	JMD
				62.5	23.3	7.5	0.8	1.7	4.2	RP
Watching a movie at a theatre	3.1	4	0.537	47.2	22.2	11.1	5.6	8.3	5.6	JMD
				48.3	16.7	14.2	1.7	6.7	12.5	RP
Using a ruler	46.0	4	0.001***	23.6	27.8	23.6	8.3	9.7	6.9	JMD
				70.0	14.2	5.8	2.5	0.8	6.7	RP
Walking outdoors during the day	2.3	4	0.682	68.1	20.8	8.3	1.4	1.4	0.0	JMD
				73.3	16.7	8.3	0.0	1.7	0.0	RP
Preparing meals	8.7	2	0.013**	59.7	29.2	8.3	0.0	0.0	2.8	JMD
				78.3	14.2	4.2	0.0	0.0	3.3	RP
Grooming	5.4	3	0.144	76.4	20.8	2.8	0.0	0.0	0.0	JMD
				85.0	10.0	3.3	1.7	0.0	0.0	RP
Using a screwdriver	23.3	4	0.001***	47.2	26.4	15.3	1.4	1.4	8.3	JMD
				70.0	11.7	1.7	5.0	0.8	10.8	RP
Using escalators	6.2	3	0.104	86.1	8.3	1.4	0.0	4.2	0.0	JMD
				70.0	19.2	4.2	0.0	5.0	1.7	RP
Watching television	40.0	4	0.001***	36.1	31.9	20.8	9.7	1.4	0.0	JMD
				80.0	12.5	6.7	0.8	0.0	0.0	RP
Grocery shopping	15.3	4	0.004***	27.8	36.1	13.9	5.6	6.9	9.7	JMD
				56.7	20.8	9.2	1.7	6.7	5.0	RP
Using public transport	11.9	4	0.018**	25.0	26.4	15.3	8.3	2.8	22.2	JMD
				40.8	15.0	9.2	1.7	1.7	31.7	RP

*JMD = juvenile macular dystrophy; RP = retinitis pigmentosa.
**$0.01<p\leq0.05$.
***$p\leq0.01$.

between visual acuities and areas of central scotoma to the II-4-e target ($r(69)=-0.40$, $p \leq 0.01$). Across categories of questions, the correlations were similar for both visual acuity and area of central scotoma to the II-4-e target. In only seven out of 33 cases were the two vision indexes not compatible. These relations were reflected in the pattern of correlations between the patients' responses to the questionnaire items and the vision measures.

The juvenile macular dystrophy patients' responses to a majority of the questionnaire items (24 out of 33) were significantly correlated with the clinical vision measures.

Not surprisingly, this was true for the category "activities involving central vision," where the correlations in Table 1 indicate that the patient responses were correlated with either their visual acuities or the areas of central scotoma to the II-4-e target, or both, with the exception of one item. Only eight items from the remaining categories were not related to the clinical vision measures.

We then compared the response patterns of the patients with juvenile macular dystrophies and those with RP using a χ^2 analysis. The χ^2 values, degrees of freedom, and the significance values for these analyses are given in Table 2. As indicated by the asterisks showing statistical significance, there were clear differences in the response patterns of the two groups in five of the six functional categories. Twenty five items showed statistically significant differences between the two groups, and the responses were primarily of two types. There were tasks which the patients with juvenile macular dystrophies reported as being more difficult, and other tasks which the patients with RP reported being more difficult. The items that were reported as more difficult by the patients with juvenile macular dystrophies included every item in the "activities involving central vision" and "miscellaneous" categories, and the activity "finding particular items in a store" in the "mobility" category. By contrast, the items that were reported as more difficult for the patients with RP included every item in the "mobility" category except "finding particular items in a store," and both items within the category "negotiating steps". "Driving" was a category which had the lowest participation so it was difficult to compare these items in this category with the items from other categories. However, both groups reported some degree of difficulty with items in this category.

There were no statistically significant differences between the groups for eight of the 33 items. Five of these eight items were not correlated with the clinical vision measures for the patients with juvenile macular dystrophies, and included the items "participating in social gatherings," "driving at night," "eating meals," "grooming," and "using escalators." In the case of the remaining three items, "engaging in physical exercise," "watching a movie at a theatre," and "walking outdoors during the day," a majority in both groups reported having no difficulty with these activities.

Discussion

From the correlational analyses, we determined that there were three types of activities within our questionnaire: (1) those that showed a correlation with both visual acuity and area of central scotoma (17 items); (2) those that were correlated with either visual acuity or area of central scotoma (seven items); and (3) those that were not correlated with either clinical vision measure (nine items). The majority of the items (17 out of 33) fell into the first type. Three items—"watching television," "walking through shopping malls," and "walking outdoors during the day"—correlated with the visual field, but not the visual acuity measures. This finding has some face validity in that these three activities appear to represent tasks that would involve more extrafoveal than foveal function. Four items—"reading ingredients on cans of food," "finding particular items in a store," "driving in unfamiliar areas," and "engaging in physical exercise"—were correlated with visual acuity, but not with the visual field measures.

Nine items fell into the third type and did not show any correlation with the vision measures for the juvenile macular dystrophies group. Of interest, five of the nine items ("participating in social gatherings," "driving at night," "eating meals," "grooming," "using escalators") also did not discriminate between the two patient groups in the χ^2 analysis. Of the remaining four items which did not show any correlation, three items ("preparing meals," "using a screwdriver," and "finding destination in unfamiliar building") were relatively easy activities for both groups, and one item ("driving during the day") was an activity that fewer individuals participated in from either group. Three additional items ("engaging in physical exercise," "watching a movie at a theatre," and "walking outdoors during the day") which did not discriminate between the two patient groups, but were correlated with the vision measures, were all activities for which a large percentage of both groups reported having no difficulty. Overall, there were 12 items out of 33 which did not show correlations with visual function and/or did not discriminate between the two patient groups. These 12 items appear to have marginal usefulness in assessing visual function.

The 21 items that showed a correlation with the clinical vision measures and discriminated between the two groups represent a broad spectrum of activities in which a large number of individuals participated and may represent a set of questions that is effective at measuring self reported visual function. As was described in our earlier study,[2] the items were drawn from a variety of sources including the Activities of Daily Vision Scale (ADVS) and the Functional Assessment Self Report Inventory of Szlyk *et al*,[1] and we composed additional questions intended to target potentially problematic activities for patients with RP. The ADVS and the Functional Assessment Self Report Inventory have both been used to assess patients with central vision loss[1 4 10] as well as peripheral vision loss.[1 11] Isfahani *et al* [11] found that the severity of visual field defects in glaucoma patients correlated with impairments of daily activities as measured with the ADVS that were independent of visual acuity. It was our intention to produce a questionnaire that would include items which would represent both central and peripheral challenges. In future versions of the questionnaire, it may be useful to eliminate those items such as "eating meals" and "grooming," for example, which neither correlated with clinical measurements of visual function, nor demonstrated significantly different patterns of response between the patient groups.

In a recent study involving only patients with RP who had a wide range of visual acuity loss,[12 13] we compared the patients' self reported difficulty with the reports of verifiers (for example, spouses, relatives, siblings of the patients), and with the actual performance of the task by the patient as assessed by a certified orientation and mobility specialist. The patients' reports were highly significantly correlated with the reports of the verifiers, and with the assessments by the orientation and mobility specialist. Regardless of the measure used, patients' performance overall showed more significant correlations with the measures of central vision, visual acuity, and contrast sensitivity, than with visual field or electroretinogram measures. Because most tasks performed in daily life involve central vision, it is not surprising that in the present study the patients with poor central vision, but normal peripheral visual fields, would report relatively more difficulties in functioning compared with the patients with RP, who had visual acuities of 20/40 or better with peripheral visual field loss.

Some activities clearly differentiated between the two groups in the present study and some showed no difference. Our results provide insight into the types of perceived difficulties in performing tasks of everyday life in patients with these disorders. Since the Americans with Disabilities Act was passed in 1990, interest has been generated in determining the functional limitations imposed by various types of visual impairments. From a clinical perspective, to be more sensitive to the individual needs of patients with vision loss, it is important to be aware of the varying profiles of functional impairment produced by different visual disorders.

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