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► **To cite this version:**

Gilles C Martin, Matthieu Robert, Georges Challe, Nhung Th Trinh, Tania Attie-Bitach, et al.. Functional Vision Analysis in Patients With CHARGE Syndrome. *Journal of Pediatric Ophthalmology and Strabismus*, 2020, 57 (2), pp.120-128. 10.3928/01913913-20200207-02 . hal-02995263

HAL Id: hal-02995263

<https://hal.sorbonne-universite.fr/hal-02995263v1>

Submitted on 9 Nov 2020

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1 Functional vision analysis in a series of patients with CHARGE syndrome

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36

37 Word count: 2912/4000

38 Acknowledgments:

39 We thank all the patients and families for their participation. We are thankful to Ms Wiam
40 BHIA for her help in collecting consent forms, to Prof. Martin CHALUMEAU for his
41 review of the manuscript, and to Ms Charlotte CREUX for her precious help in creating the
42 questionnaire. We thank Ms Laura SMALES for helping with editing and English writing.

43 Abstract:

44 **Purpose:** CHARGE syndrome (CS) is a multiple malformative syndrome in which ocular
45 colobomas cause visual impairment. Data are lacking regarding visual function because
46 classical methods for visual acuity (VA) assessment are often not applicable in patients with
47 CS. We evaluated vision in a pilot study of patients with CS by using a new questionnaire
48 entitled VISIOCHARGE.

49 **Methods:** Ophthalmological data including fundus description and VA, when available,
50 were extracted from charts of 83 patients with CS, and VISIOCHARGE was prospectively
51 sent to 55 of them. The answers of the 36 responders (18 males) allowed for calculating 3
52 scores assessing distance-vision, near-vision, and an “overall ability” score.

53 **Results:** Visual acuity measurements were extracted from the charts of 20 of the 36 patients.
54 The mean VA was 20/50. The mean distance-vision score of 0.62 (SD 0.30) and near-vision
55 score of 0.78 (SD 0.23) were correlated with VA in the 20 patients ($\rho=0.64$, $p=0.002$ and
56 $\rho=0.61$, $p=0.005$, respectively) and were associated with the severity of the colobomatous
57 malformation ($p=0.049$ and $p=0.008$, respectively). Severity of the ocular malformation was
58 not associated with overall ability score ($p=0.64$).

59 **Conclusions:** VISIOCHARGE is feasible in patients with CS and may help in the
60 assessment of visual function. The mean VA and the answers to the questionnaire showed
61 relatively good visual skills in these patients in everyday life, even in those with bilateral
62 colobomas, which contrasts with the pessimistic conclusions usually resulting from the
63 initial fundus examination.

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65 Number of words: 238/250

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69 **INTRODUCTION**

70 CHARGE syndrome (CS) is a multiple malformative syndrome, with coloboma representing
71 a major feature and diagnostic criterion^{1,2}. This ocular anomaly is found in 72% to 95% of
72 patients with CS³⁻⁸ and represents an important cause of disability in affected patients.
73 However, previous studies mainly focused on the anatomical aspects of the ocular
74 manifestation of the syndrome and rarely reported results from visual acuity (VA) and/or
75 visual function assessment. This lack of data is due in part to the difficulty in obtaining a
76 reliable VA value in many patients with CS, as classical ophthalmological evaluation
77 methods are often not applicable because of the multiple sensory deficits and/or cognitive
78 disabilities of such patients. The few series reporting VA in CHARGE syndrome found
79 mean values below 20/60⁴⁻⁸. However, in our experience, the parents, relatives and
80 professionals caring for these patients often report good visual skills in everyday life, which
81 contrasts with the often pessimistic conclusions from the ophthalmological examinations, in
82 particular neonatal examinations. This contrast points out the need for a new tool to assess
83 the real visual function in patients with CS regardless of the severity of their sensory, motor
84 and/or cognitive impairments.

85 In this study, we aimed to develop an original self-administered questionnaire designed for
86 patients with CS (VISIOCHARGE) and we used it to evaluate the functional vision of
87 patients with CS. The other goals were to describe the ocular features of a large series of
88 patients with CS, to analyze the links between the severity of the ocular malformation and
89 practical visual function and to confirm the previously suspected correlation between visual
90 impairment and poor developmental milestones in these children^{9,10}.

91

92

93 PATIENTS & METHODS

94 Questionnaire development

95 The questionnaire, entitled VISIOCHARGE (see Appendix) was built from observations and
96 comments of pediatricians, ophthalmologists and orthoptists used to following up patients
97 with CS. A first version of the questionnaire was randomly sent to 4 families to obtain their
98 comments on its quality, feasibility and understandability. After a review, we adopted a final
99 version of the questionnaire consisting of 30 items in 3 categories: 1) parental evaluation of
100 global vision (2 items), designed to assess parents' feeling about the importance of the visual
101 impairment of their child and its consequences on everyday life; 2) evaluation of distance
102 vision (9 items); and 3) evaluation of near vision (10 items). Additional questions were
103 asked to evaluate the ophthalmological follow-up (4 items), educational level and age of
104 walking acquisition of the patient (5 items). Some free space was available at the end of the
105 document for open answers and comments.

106

107 Study population

108 All patients with CS followed or seen at least once between January 1, 1990 and December
109 31, 2016 in the pediatric and/or ophthalmologic departments of Necker-Enfants Malades
110 Hospital and/or La Pitié-Salpêtrière Hospital were included. CS diagnosis was established
111 clinically by using the Blake and Verloes criteria^{1,2}. When available, their CHD7 mutation
112 status was extracted from medical charts. The VISIOCHARGE questionnaire was sent by
113 mail between January 1 and December 31, 2017 to the most recent address of the patient
114 registered in the hospital database, accompanied by an information letter and a consent form.
115 The patients, or their parents in case of inability to read and/or to understand the questions,
116 were asked to complete the questionnaire and return it by mail to the investigator with a
117 signed consent form. A few days after sending the questionnaire, investigators contacted the
118 patients or their parents by phone call to further explain the aim of the study and answer their

119 questions. Patients with CS who had died before the beginning of the study were excluded
120 and their parents were not asked for participation. This study was approved by the ethics
121 committee of Necker-Enfants Malades Hospital on October 31, 2016 (authorization number:
122 2016-VA 24-R1), and adhered to the tenets of the Declaration of Helsinki.

123

124 **Data collection and outcome measures**

125 The following information was extracted from medical charts of patients: the most recent
126 corrected best binocular VA, the description of fundus of each eye after dilation (presence of
127 chorioretinal and/or optic disk coloboma, involvement of the fovea and/or the optic disk
128 inside the coloboma), and presence of nystagmus, strabismus, and any other anomalies seen
129 during the ophthalmological examination, including ptosis, facial palsy, congenital cataract
130 or retinal detachment.

131 VA was measured by using the French Monoyer chart and was expressed in decimal,
132 fraction (in feet) and LogMAR units.

133 On the basis of data from the medical chart, we assigned patients to two groups based on the
134 severity of anatomical lesions. Group A included patients with no coloboma in both eyes or
135 with a coloboma in one or both eyes but at least one eye having a fully preserved optic disk
136 and fovea. Group B included patients with a coloboma involving the optic disk in both eyes,
137 with or without involvement of the fovea.

138 Answers to each item of the questionnaire were analyzed independently to detail the visual
139 ability. We then calculated three scores by giving a value in points for the different items;
140 the maximum score corresponded to the most difficult or smallest things to see. The three
141 scores calculated were a distance-vision score, a near-vision score, and an overall ability
142 score. The overall ability score combined the answers to six items exploring the ability to
143 perform six tasks: watch TV, move around indoors in a familiar place, move around indoors

144 in an unknown place, move around outdoors, use a tablet PC, use a smartphone. The scores
145 were expressed as decimals between zero and one.

146 To simply assess the psychomotor development of patients, the questionnaire asked about
147 the age of walking acquisition, which was verified in medical charts, and the most recently
148 attended educational structure. These educational structures were classified from level 1 to 5,
149 reflecting the communication and cognitive abilities of the patients: 1) in a regular school
150 without any specific assistance; 2) with an assistant dedicated to the child in a regular
151 school; 3) in a specific classroom and/or structure for deaf and/or blind children, without
152 cognitive disabilities; 4) in a structure dedicated to deaf and/or blind children with cognitive
153 impairment but communication abilities; and 5) severe cognitive disabilities without any
154 acquired language and poor learning.

155

156 **Statistics**

157 Continuous variables were expressed as mean with standard deviation (SD). Because the
158 sample was too small, we could not divide the ophthalmological gravity of patients into
159 more than two groups for analyzing the correlation between anatomic and functional data.
160 We compared groups by Kruskal Wallis, Mann-Whitney and Fisher tests, and used the
161 Spearman for correlations. Significance was considered at two-sided $p < 0.05$.

162

163 **RESULTS**

164 The database review found 83 patients (46 males) followed in the two centers for typical CS
165 with or without ophthalmological involvement. Among the 83 patients, 10 were deceased
166 and 18 were followed elsewhere. Thus, we sent the VISIOCHARGE questionnaire to 55
167 patients included in the prospective part of the study. Four questionnaires were returned
168 because of a wrong address, and 15 families received the letter but did not answer or refused
169 to take part in the study. Finally, among the 55 solicited patients, 36 (18 males) with

170 completed VISIOCHARGE questionnaires were included in the analysis (response rate =
171 66%). The mean age of responders was 13.8 years (SD 6.9). The demographic,
172 ophthalmological and neurodevelopmental characteristics of the 36 responders were
173 comparable to those of the 83 patients in the whole series (Table 1) allowing for discussion
174 and conclusions. A mutation in *CHD7* was found in 31 children of the 33 investigated
175 patients (94%). The genetics investigation had not been performed for 3 others.

176 Of the 36 patients, 32 (89%) had at least one ocular coloboma of the posterior segment
177 reported in their medical chart; 4/32 (11%) had presented a unilateral retinal detachment
178 (RD). Overall, 27/36 (75%) patients wore glasses; the mean age at the first optical correction
179 was 4 years (SD 3.7).

180

181 **Visual function and its consequences for activities of daily life**

182 Of the 36 patients, only 20 (56%) had VA data available in medical charts. In these patients,
183 the mean VA was 0.42 (20/50, 0.40 LogMAR). When we excluded the only patient without
184 coloboma with a measurable VA, the mean VA stayed similar (0.40 LogMAR). For 26/36
185 (72%) patients, their parents reported trouble related to the vision of their child; for 11/36
186 (30%), their child was declared to be “very bothered” by it (Table 2). Nevertheless, 32/36
187 (89%) patients were able to use electronic devices such as smartphones or digital tablets;
188 31/36 (86%) were able to watch television, 22/36 (61%) reported an ability to recognize a
189 familiar face at more than 2 m, and 19/36 (53%) were able to read or identify Arial
190 characters of 18 size at a reading distance of 40 cm. The mean distance-vision, near-vision,
191 and overall ability scores were 0.62 (SD 0.30), 0.78 (SD 0.23) and 0.79 (SD 0.25),
192 respectively.

193 For the 20 patients with a VA measurement, the distance-vision and near-vision scores from
194 the VISIOCHARGE questionnaire were positively correlated with measured VA ($\rho=0.64$,
195 $p=0.002$, and $\rho=0.61$, $p=0.005$, respectively). Parental evaluation of the global vision of their

196 child was correlated with the distance-vision and near-vision scores ($r_s=0.61$, $p<0.001$, and
197 $r_s=0.63$, $p<0.001$, respectively) and with measured VA ($r_s=0.68$, $p<0.001$).

198 **Developmental features**

199 In this series, the mean age of walking acquisition was 35.4 months (SD 15.0). The distance-
200 vision score was negatively correlated with age of walking acquisition ($\rho=-0.38$, $p=0.037$).
201 Age of walking acquisition was not correlated with parental evaluation of the global vision
202 of their child or near-vision or overall ability score ($r_s=-0.22$, $p=0.23$; $\rho=0.03$, $p=0.86$;
203 $\rho=0.02$, $p=0.92$, respectively). Regarding educational levels, most patients had been
204 schooled in structures of types 2, 3 and 4. The distance-vision, near-vision and overall ability
205 scores were all negatively correlated with educational level ($r_s=-0.54$, $p<0.001$; $r_s=-0.45$,
206 $p<0.001$; $r_s=-0.39$, $p=0.02$, respectively). Educational level was not correlated with parental
207 evaluation of the global vision of their child ($r_s=-0.27$, $p=0.11$).

208

209 **Correlation between anatomic and functional data**

210 Patients in group A (mild anatomical lesions, $n=18$) were compared to those in group B
211 (severe anatomical lesions, $n=17$) (Table 3). One patient was excluded from this analysis
212 because of an incomplete description of the fundus. As expected, the mean VA was
213 significantly worse for group B than group A patients: 0.23 (20/80, 0.64 LogMAR) vs 0.88
214 (20/25, 0.05 LogMAR), $p<0.001$. Similarly, the mean distance-vision and near-vision scores
215 were significantly lower for group B than group A (0.54 vs 0.71, $p=0.049$, and 0.69 vs 0.88,
216 $p=0.008$, respectively). However, the 2 groups did not differ in parental evaluation of the
217 global vision of their child ($p=0.07$) or in the overall ability score from the questionnaire
218 (0.76 vs 0.83, $p=0.64$). The two groups did not differ in developmental features. Taken
219 separately, the only two items showing a significant difference between the two groups were
220 those exploring the ability to recognize a familiar face at a given distance ($p=0.008$) and the
221 smallest size of Arial text readable ($p=0.03$).

222 **Comments from responders**

223 Ten (28%) patients (or their parents) reported better performance in near vision than distance
224 vision. For 6 (17%) patients, a photophobia was also spontaneously reported in
225 questionnaires. The parents of a young patient reported that at birth, their child had been
226 given a prognosis of severe vision, total blindness, after the first ophthalmological
227 examination because of the severity of the colobomas involving the fovea in both eyes. The
228 VA of this child was actually measured at 0.3 (20/60, 0.48 LogMAR). His distance-vision,
229 near-vision and overall ability scores were 0.70, 0.60 and 1, respectively. One patient
230 spontaneously highlighted a prosopagnosia (difficulty in recognizing faces out of their
231 context).

232

233 **DISCUSSION**

234 This series is one of the largest to describe the ocular features in CS, and the first to
235 specifically address the question of visual function, showing better visual skills than
236 previously reported. The most recent studies^{4,5,7} reported VA under 0.3 (20/60, 0.48
237 LogMAR) in 58% to 67% of patients with CS for whom VA was measurable, which
238 contrasts with the 30% in our series, closer to the 17% found by Russel-Eggitt *et al* (6).
239 However, these comparisons are difficult to interpret because of the very small proportion of
240 patients in each series for whom VA was measurable. With 20 (56%) patients having a
241 measured VA, our study is the first to report such a high proportion of available data.
242 However, the substantial number of missing data is also the main reason that led us to look
243 for another way to explore visual function in this specific population.

244 In the ophthalmological field, some examination methods exist to assess visual function in
245 non-verbal children who are very young or have intellectual impairment. Among them, the
246 preferential looking procedures¹¹ (Cardiff Acuity Test, Teller Acuity Test), optokinetic
247 nystagmus assessment, and visual evoked potentials are the most commonly used. However,

248 these tools are not widely available in routine care and may not be suitable for some patients
249 with CS because of cognitive disabilities reducing their attention or some ophthalmological
250 features such as oculomotor palsies or nystagmus, commonly found in this population.
251 Questionnaires assessing functional vision in the pediatric field are being increasingly used
252 to assess the consequences of the visual impairment on quality of life. We reviewed seven
253 available questionnaires¹²⁻¹⁸ but none were suitable for patients with CS because the
254 questions implied an absence of sensory, motor or cognitive disabilities besides the visual
255 impairment (Table 4). We found two questionnaires designed for children with
256 disabilities^{19,20}. However, one was not appropriate for regular self-administered evaluation,
257 because some items seemed equivocal, whereas the other was specifically aimed at children
258 with severe cognitive disabilities, which is not a systematic feature of patients with CS.
259 Thus, we aimed to develop an original questionnaire, suitable for every patient, regardless of
260 the age or any motor and/or cognitive disability. VISIOCHARGE not only proved is
261 feasibility in a heterogenous population of patients with CS, but also demonstrated that most
262 of the patients with CS were able to perform similar tasks as other children. Most of the
263 patients were not very bothered by their visual impairment, especially in tasks involving near
264 vision. The main difficulties reported were in distance vision and outdoor activities.
265 VISIOCHARGE could reflect visual function because the distance-vision and near-vision
266 scores were well correlated with VA, when available. To confirm these results, this
267 questionnaire should be tested in other diseases featuring impaired visual function in
268 children with or without associated disabilities. Indeed, the main limitation of this study is
269 that this questionnaire has not been rigorously validated yet. Now that we have proven the
270 questionnaire's feasibility on a pilot population, its use in a much larger population is
271 possible, and its validation by evaluating its metrological qualities has to be done. Another
272 important limitation of the study is the risk of subjectivity, unavoidable with self-
273 administered questionnaires and perhaps more so when a relative performs it. However,

274 because we wanted to assess visual skills in everyday life, we cannot totally suppress this
275 subjectivity.

276 With a total of 83 charts reviewed, our series is the largest to detail the ocular features in CS.
277 As previously described, the most frequent feature is chorio-retinal coloboma, found in 83%
278 of our patients, close to the 82%, 79% and 90% of Russel-Eggit *et al.*, Tellier *et al.* and
279 Strömland *et al.*, respectively⁶⁻⁸. Retinal detachment was found in 4,4% of colobomatous
280 eyes, in the low average of previously published data about RD complicating colobomas,
281 suggesting that CS may not be a risk factor for RD.

282 The correlation between anatomy and function is incomplete. Although VA and distance-
283 vision and near-vision scores were better in children with peripheral colobomas than those
284 with colobomatous lesions involving both the optic nerve and/or macula, the results from our
285 questionnaire did not find any difference between these patients in overall ability score and
286 parental evaluation of their global vision. This lack of correlation may be due in part to a
287 potential failure of our questionnaire to reveal a difference. However, these results confirm
288 the general impression of parents and caregivers that children with CS and large colobomas
289 can develop some surprising compensatory strategies, allowing them to use their remaining
290 vision, either central or peripheral. Another explanation for the lack of anatomic and
291 functional correlation is the difficulty for ophthalmologists to assess macular anatomy by
292 fundus examination alone, as it was previously shown²¹. Thus, as suggested by Nishina *et*
293 *al.*, it seems crucial not to predict poor vision in a neonate with CS and bilateral coloboma
294 because our experience showed that some of these children can later show correct visual
295 function⁵.

296 Our series also confirmed the previously suspected association between visual function and
297 some developmental parameters^{9,10} independent of other manifestations of the syndrome.
298 Late acquisition of walking has been found negatively correlated with distance-vision score,
299 and educational level has also been correlated with all visual function scores. This

300 observation reinforces the promotion of regular and rigorous ophthalmological care for every
301 patient with CS, regardless of the severity of the ocular malformation, to assure the best
302 visual prognosis as well as good general development.

303 Assessment of visual function in patients with CS may be challenging because VA
304 measurements are often not possible in these patients. We present encouraging preliminary
305 results with an original questionnaire, specifically designed for children with visual
306 impairment and associated sensory, motor and/or cognitive disabilities. The relatively good
307 visual skills of the patients in this series contrast with the often-pessimistic conclusions from
308 initial ophthalmological examinations. Ophthalmologists should not give a poor visual
309 prognosis to parents of a newborn recently diagnosed with CS and bilateral coloboma. They
310 should encourage parents to stimulate their child with lights and colored objects as much as
311 possible to foster the developmental of their social brain and visual cortex.

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361 nerve involvement, size, or the fovea? *Ophthalmology* 1997; 104 : 1367-1368.

363 **Table 1: Comparison of the 36 responders to the VISIOCHARGE questionnaire and**
 364 **the total number of CHARGE syndrome patients followed in our center**
 365

	Charts reviewed n=83	VISIOCHARGE responders n=36	p
Sex, n patients (%)			0.69
Male	46 (55)	18 (50)	
Female	37 (45)	18 (50)	
Ocular features, n patients (%)			
Posterior coloboma	69 (83)	32 (89)	0.58
- unilateral	14 (17)	7 (19)	
- bilateral	55 (66)	25 (69)	
Iris coloboma	12 (14)	7 (19)	0.59
- unilateral	6 (7)	3 (8)	
- bilateral	6 (7)	4 (11)	
Microphthalmos	28 (34)	15 (42)	0.41
- unilateral	25 (30)	13 (36)	
- bilateral	3 (4)	2 (6)	
Ptosis	14 (17)	7 (19)	0.80
Nystagmus	24 (29)	11 (31)	1
Facial palsy	24 (29)	10 (28)	1
Congenital cataract	4 (5)	1 (3)	1
- unilateral	3 (4)	1 (3)	
- bilateral	1 (1)	0	
Retinal detachment	6 (7)	4 (11)	0.49
- unilateral	6 (7)	4 (11)	
- bilateral	0	0	
Severity of the coloboma*			0.55
Group A, n patients (%)	42 (51)	18 (50)	
Group B, n patients (%)	36 (43)	17 (47)	
Unclassified	5 (6)	1 (3)	
Age of walking (months)			
Mean (SD)	36.9 (15.7)	35.4 (15.0)	0.66
Min-max	19-78	19-78	
Visual acuity (decimal):			
Mean	0.41	0.42	0.97
Unmeasurable	48 (58)	16 (44)	

366 *Group A, patients with no coloboma in both eyes or with coloboma in one or both eyes but at least
 367 one eye having a fully preserved optic disk and fovea; Group B, patients with coloboma involving
 368 the optic disk in both eyes, with or without involvement of the fovea.
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Table 2: Answers for the VISIOCHARGE questionnaire for the 36 children

Global vision	Parental evaluation of the global vision of their child, n (%)	
	Normal, or abnormal but without inconvenience	10 (28)
	Slightly bothered by his/her visual impairment	7 (19)
	Moderately bothered by his/her visual impairment	8 (22)
	Very bothered by his/her visual impairment	11 (31)
Distance vision	My child can watch TV, n (%), IDK*	31 (86), 0
	If yes, the TV is placed at a distance of: n (%)	
	> 2 m	7/31 (23)
	50 cm to 2 m	17/31 (54)
	< 50 cm	7/31 (23)
	My child can recognize a familiar face at a maximum distance of: n (%)	
	> 10 m	9 (25)
	2 m to 10 m	13 (36)
	< 2 m	12 (33)
	IDK	2 (6)
	When looking at the sky, my child can see:	
	The moon, at night, n (%), IDK*	19 (53), 9
A plane, at daytime, n (%), IDK*	19 (53), 5	
The visual impairment of my child bothers him/her in moving:		
Indoor, in a familiar place, n (%), IDK*	3 (8), 4	
Indoor, in an unknown place, n (%), IDK*	9 (25), 5	
Outdoor, n (%), IDK*	19 (53), 2	
Distance vision score, mean (SD)	0.62 (0.30)	
Near vision	My child can use a tablet PC, n (%), IDK*	32 (89), 0
	If yes, he uses it:	
	In "normal" conditions	28/32 (88)
	With specific adjustments (e.g., character magnification)	4/32 (12)
	My child can use a smartphone, n (%), IDK*	32 (89), 1
	My child can read a text (or identify drawings) of a minimum size of (at a distance of 40 cm): n (%)	
	Arial 8	12 (33)
	Arial 10	4 (11)
	Arial 18	3 (8)
	Arial 28	4 (11)
Arial 48	6 (17)	
> Arial 48	2 (6)	
My child cannot perform this test	5 (14)	
During lunch time, my child can see on the table in front of him/her: n (%)		
A grain of rice	29 (81)	
An olive	35 (97)	
An apricot/plum/strawberry	35 (97)	
An apple/orange	36 (100)	
My child is able to see and catch a strand of hair, n (%), IDK*	22 (61), 2	
Near vision score, mean (SD)	0,78 (0.23)	
Overall ability score (TV, motion, smartphone, tablet PC), mean (SD)	0,79 (0.25)	

373 * IDK: I don't know

374

375 **Table 3: Comparison of visual and developmental features by severity of the**
 376 **colobomatous malformation: Group A and B patients**
 377

	Group A* (n=18)	Group B* (n=17)	p
Age (years), mean (SD)	13.1 (6.8)	14.7 (7.3)	0.50
Parental evaluation of the overall vision of their child, n (%)			0.07
Normal, or abnormal but without inconvenience	8 (44)	2 (12)	
Slightly bothered by his/her visual impairment	2 (11)	5 (29)	
Moderately bothered by his/her visual impairment	5 (28)	3 (18)	
Very bothered by his/her visual impairment	3 (17)	7 (41)	
Visual acuity (LogMar), mean (SD)	0.05 (0.10)	0.64 (0.28)	<0.001
Distance-vision score, mean (SD)	0.71 (0.31)	0.54 (0.27)	0.049
Near-vision score, mean (SD)	0.88 (0.20)	0.69 (0.22)	0.008
Overall ability score, mean (SD)	0.83 (0.20)	0.76 (0.30)	0.64
Age of walking (months), mean (SD)	34.7 (15.9)	36.2 (15.1)	0.79

378 *Group A, no coloboma in both eyes or with a coloboma in one or both eyes but at least one eye
 379 having a fully preserved optic disk and fovea; Group B, a coloboma involving the optic disk in both
 380 eyes, with or without involvement of the fovea.
 381
 382

383 **Table 4: Comparison of existing questionnaires evaluating visual function and visual**
 384 **disability in children**
 385

	Age range (years)	Target children	Person interviewed	No. of items	Evaluated parameters
VAS ^a	2–18	Children with visual impairment with no other sensorial, physical or cognitive disability	Parents	16	Visual acuity, visual field, color vision
CVFQ ^b	≤ 7	Children with visual impairment with or without developmental delay	Parents	40	Personal, social and familial impact of visual disability
CVAQC ^c	5–18	Children with acquired spoken language and no sensorial, physical or cognitive disability	Child	25	Ability to complete tasks requiring vision
IVI_C ^d	8–18	Children with visual impairment with acquired spoken language and no other sensorial, physical or cognitive disability	Child	24	Personal, social and scholar impact of visual disability
LVP_FVQ II ^e	8–18	Children in developing countries with acquired spoken language and no sensorial, physical or cognitive disability	Child	23	Grading of visual impairment
FVQ_CYP ^f	10–15	Children with acquired spoken language and no sensorial, physical or cognitive disability	Child	36	Ability to complete tasks requiring vision
PreViAs ^g	≤ 2	Babies, except premature infants, without a “major” adverse medical history or developmental delay	Parents	30	Reactions of the infant to visual stimulations

386 ^aVisual Ability Score (Katsumi *et al.*, 1998)

387 ^bChildren’s Visual Function Questionnaire (Felius *et al.*, 2004)

388 ^cCardiff Visual Ability Questionnaire for Children (Khadka *et al.*, 2010)

389 ^dImpact of Vision Impairment for Children (Cochrane *et al.*, 2011)

390 ^eL.V. Prasad – Functional Vision Questionnaire (Gothwal *et al.*, 2012)

391 ^fFunctional Vision Questionnaire for Children and Young People (Tadic *et al.*, 2013)

392 ^gPreverbal Visual Assessment questionnaire (Pueyo *et al.*, 2014)

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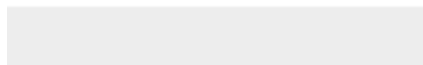
395 Appendix: The VISIOCHARGE Questionnaire (translated from its original French version)

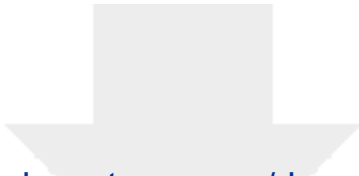


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Supplemental Material / Data

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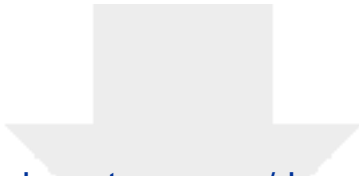
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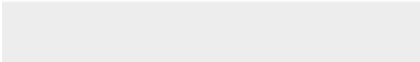
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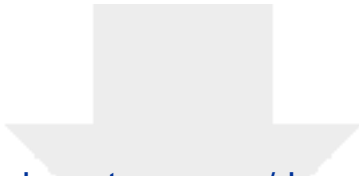
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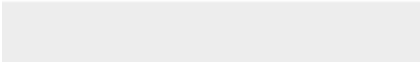
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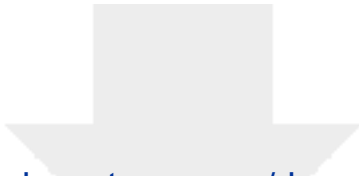




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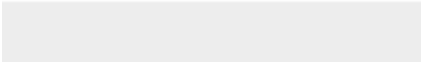
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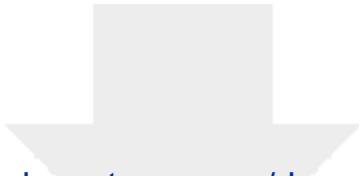




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