Title

Development of novel patient-reported outcome (PRO) and observer-reported outcome (ObsRO) instruments in Retinitis Pigmentosa (RP) and Leber Congenital Amaurosis (LCA): ViSIO-PRO and ViSIO-ObsRO

Authors

Isabelle Audo[1], Francesco Patalano[2] (ORCID: 0000-0001-7015-6084), Christel Naujoks[2] (ORCID: 0000-0002-3799-6521), Claudio Spera[2], M. Dominik Fischer[3-5], Jane Green[6], Christine Kay[7], Todd Durham[8] (ORCID: 0000-0002-0363-0329), Nicola Williamson[9], Helena Bradley[9], Melissa Barclay[9], Kieran Boparai[9], Judit Banhazi[2]

Affiliations

[1]Sorbonne Université, INSERM, CNRS, Institut de la Vision, CHNO des Quinze-Vingts, National Rare Disease Center REFERET and INSERM-DGOS CIC 1423, F-75012 Paris, France;

[2]Novartis Pharma AG, Basel, Switzerland;

[3]University of Oxford, Department of Clinical Neurosciences, Oxford, UK

[4]Oxford Eye Hospital, Oxford University Hospitals NHS Foundation Trust, Oxford, UK

[5]Centre for Ophthalmology, University Hospital Tuebingen, Tuebingen, Germany;

[6]Discipline of Genetics, Faculty of Medicine, Memorial University of Newfoundland, Craig L Dobbin Genetics Research Centre, St. John's, Canada;

[7]Vitreoretinal Associates, Gainesville, Florida, USA;

[8]Foundation Fighting Blindness, Columbia, Maryland, USA;

[9]Adelphi Values Ltd, Cheshire, UK.

Corresponding Author: Helena Bradley, helena.bradley@adelphivalues.com

27 **Supplementary material**

Supplementary Table 1. Ovid literature search strategy*

Search type	Search terms
Disease-related terms	Bothnia retinal dystrophy OR fundus albipunctatus OR newfoundland rod-cone dystrophy OR retinitis punctata albescens AND retinitis pigmentosa
Concepts of interest	quality of life OR disability OR morbidity OR mortality OR epidemiology OR prevalence OR incidence OR risk factor OR co-morbid OR pattern OR trend OR estimate OR project OR extrapolate OR hospitalisation OR drug utility OR resource utility OR length of stay OR emergency room OR emergency department OR economic OR cost OR expense OR work OR productive OR present OR absent OR loss
Qualitative research terms	visual activities questionnaire AND patient reported outcome OR Quality adjusted life years OR Disability adjusted life years OR preference OR questionnaire OR scale OR score OR satisfaction OR utility

**Limits applied (English, human)*

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Supplementary Table 2. Search to identify qualitative literature in the RP/LCA population

Search type	Search terms
Disease-specific terms	retinitis pigmentosa OR RPGR OR retinitis pigmentosa GTPase regulator OR bothnia dystrophy OR newfoundland rod-cone dystrophy OR *RLBP1* RP OR *RLBP1* retinitis pigmentosa OR *RLBP1* OR *RPE65*RP OR RPE65-mediated inherited retinal dystrophy OR RPE65-mediated inherited retinal disease OR RPE65-mediated inherited retinal degeneration OR RPE65-mediated inherited retinal disorder OR usher syndrome OR bardet-biedl syndrome OR laurence-moon syndrome OR laurence-moon-bardet-biedl syndrome OR

Supplementary Table 2. Search to identify qualitative literature in the RP/LCA population

Search type	Search terms
	bassen-kornzweig syndrome OR refsum disease OR refsum syndrome OR NARP syndrome OR neuropathy ataxia retinitis pigmentosa OR leber congenital amaurosis OR alstrom syndrome OR early childhood-onset retinitis pigmentosa OR severe early childhood onset retinal dystrophy OR early childhood onset severe retinal dystrophy
Qualitative research terms	qualitative OR hermeneutic OR interview OR delphi OR focus group OR phenomenology OR ethnography OR grounded theory OR thematic analysis AND exp OR qualitative research OR qualitative analysis OR exp interview OR interviewers OR interviewing OR interviews as topic OR exp focus groups OR exp phenomenology OR ethnography OR grounded theory OR thematic analysis

Supplementary Table 3. Publications used in RLBP1 RP qualitative and instrument review

Publication
Sloane, M. E., Ball, K., Owsley, C., Bruni, J. R. and Roenker, D. L. (1992) The Visual Activities Questionnaire: Developing an instrument for assessing problems in everyday visual tasks. In Paper Presented to the Topical meeting of the Optical Society of America, America.
Maw, M. A., Kennedy, B., Knight, A., Bridges, R., Roth, K. E., Mani, E. J., et al. (1997) Mutation of the gene encoding cellular retinaldehyde-binding protein in autosomal recessive retinitis pigmentosa. Nat Genet, 17(2), pp. 198-200.
Mills, R. P. (1998) Correlation of quality of life with clinical symptoms and signs at the time of glaucoma diagnosis. Trans Am Ophthalmol Soc, 96, pp. 753-812.
Botelho, P. J., Blinder, K. J. and Shahinfar, S. (1999) Familial occurrence of retinitis punctata albescens and congenital sensorineural deafness. Am J Ophthalmol, 128(2), pp. 246-7.
Burstedt, M. S., Sandgren, O., Holmgren, G. and Forsman-Semb, K. (1999) Bothnia dystrophy caused by mutations in the cellular retinaldehyde-binding protein gene (RLBP1) on chromosome 15q26. Invest Ophthalmol Vis Sci, 40(5), pp. 995-1000.
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Friedburg, C., Serey, L., Sharpe, L. T., Trauzettel-Klosinski, S. and Zrenner, E. (1999) Evaluation of the Night Vision Spectacles on patients with impaired night vision. Graefes Arch Clin Exp Ophthalmol, 237(2), pp. 125-36.
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Eichers, E. R., Green, J. S., Stockton, D. W., Jackman, C. S., Whelan, J., McNamara, J. A., et al. (2002) Newfoundland rod-cone dystrophy, an early-onset retinal dystrophy, is caused by splice-junction mutations in RLBP1. Am J Hum Genet, 70(4), pp. 955-64.

Khairallah, M., Messaoud, R., Zaouali, S., Ben Yahia, S., Ladjimi, A. and Jenzri, S. (2002) Posterior segment changes associated with posterior microphthalmos. Ophthalmology, 109(3), pp. 569-74.

Brown, M. M., Brown, G. C., Sharma, S. and Busbee, B. (2003) Quality of life associated with visual loss: a time tradeoff utility analysis comparison with medical health states. Ophthalmology, 110(6), pp. 1076-81.

Demirci, F. Y., Rigatti, B. W., Mah, T. S. and Gorin, M. B. (2004) A novel compound heterozygous mutation in the cellular retinaldehyde-binding protein gene (RLBP1) in a patient with retinitis punctata albescens. Am J Ophthalmol, 138(1), pp. 171-3.

Fishman, G. A., Roberts, M. F., Derlacki, D. J., Grimsby, J. L., Yamamoto, H., Sharon, D., et al. (2004) Novel mutations in the cellular retinaldehyde-binding protein gene (RLBP1) associated with

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Chacon-Lopez, H., Pelayo, F. J., Lopez-Justicia, M. D., Morillas, C. A., Urena, R., Chacon-Medina, A., et al. (2013) Visual training and emotional state of people with retinitis pigmentosa. J Rehabil Res Dev, 50(8), pp. 1157-68.

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Supplementary Table 4. Publications selected for full-text review in the RP/LCA population

Publication
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Simarasl Y. Challenges of Retinitis Pigmentosa (RP) Patients in Iran and Sweden (A Comparative Study). 2012.
Figueiredo MZ, Chiari BM, Goulart BN. Communication in deafblind adults with Usher syndrome: retrospective observational study. *CoDAS.* 2013;25(4):319-324.
Ming ST. RP Doesn't Mean Loss of All. Paper presented at: Retina International World Congress 2016; Taipei Taiwan.
Piscopo A, Roborel De Climens A, Brun-Strang C, Buggage R, Barbosa CD, Knoble N. Living with Usher's syndrome Type 1: Development of a disease impact model from qualitative interviews with patients and their parents. *Investigative Ophthalmology and Visual Science Conference.* 2017;58(8).
Senthil MP, Khadka J, Gilhotra JS, Simon S, Pesudovs K. Exploring the quality of life issues in people with retinal diseases: a qualitative study. *Journal of patient-reported outcomes.* 2017a;1(1):15.

Senthil M, P., Khadka J, Pesudovs K. Seeing through their eyes: Lived experiences of people with retinitis pigmentosa. *Eye (Basingstoke).* 2017b;31(5):741-748.