

What is the Wider Carer and Family Burden in Leber Hereditary Optic Neuropathy? A Mixed-Methods Study

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INTRODUCTION

- Leber Hereditary Optic Neuropathy (LHON) is a maternally inherited mitochondrial disease which leads to central vision loss due to optic neuropathy [1,2].
- Visions loss starts with a painless blurring and clouding of the central vision, typically in one eye at first, that rapidly progresses to blindness in both eyes [3].
- The condition typically presents in young adulthood [4] and causes significant impacts on many aspects of patients' lives including daily activities, emotional functioning, relationships, studies, work, recreation and finances [5].
- As such, the condition can be difficult to adjust to which anecdotally leads to substantial impacts on informal carers and family members.

OBJECTIVE

This study was designed to describe the burden of LHON for informal carers and family members using qualitative and quantitative methods.

METHODS

- Informal carers and family members (N=9, aged 18+) of patients diagnosed with LHON completed in-depth interviews.
- Participants were recruited through a patient advocacy group (PAG) in the United Kingdom (UK) and the study sponsor.
- Interviews explored how daily activities were affected as well as physical, social, emotional, work, educational and financial impacts.
- Participants completed the EQ-5D-5L, CarerQoL-7D, Work Productivity and Activity Impairment (WPAI) Questionnaire and a background questionnaire containing socio-demographic and clinical questions relating to themselves and the person they cared for.
- The study protocol was reviewed by an Independent Review Board. All study participants provided informed consent.
- Socio-demographic, clinical and HRQoL data were summarised using descriptive statistics.
- Utility scores for the EQ-5D-5L were calculated using the Hernandez et al. (2020) mapping function.
- Six scores for the CarerQoL-7D were derived using preference weights generated for the UK [6].
- Seven domain scores for the WPAI were generated using scoring algorithms [7].
- Interview data were analysed using content analysis.

QUANTITATIVE RESULTS

- Nine informal carers and family members in the UK participated in the study (age range 30-70; 8 females; 5 parents).
- Of the care recipients, n=6 were male (mean age 32, range 17-73), n=7 had the ND4 mutation, n=1 had the ND6 mutation (n=1 unknown), and all had received treatment in the past (n=6 ldebenone, n=1 GS010, n=3 other).
- **EQ-5D-5L:** The mean EQ-5D-5L utility value was 0.893 (SD= 0.245, range 0.244 – 0.988) and the mean EQ-VAS score was 84.2 (SD=8.5, range 72.5 – 95.0), see **Figure 1**.
- Seven of the nine participants responded that they had no problems in any of the EQ-5D-5L domains.
- **CarerQoL-7D:** The mean CarerQoL-7D utility was 78.4 (SD= 21.0, range 31.9 – 96.9) and the mean CarerQoL VAS score was 6.67 out of 10 (SD= 3.12, range 0.00 - 9.00), see **Figure 1**.

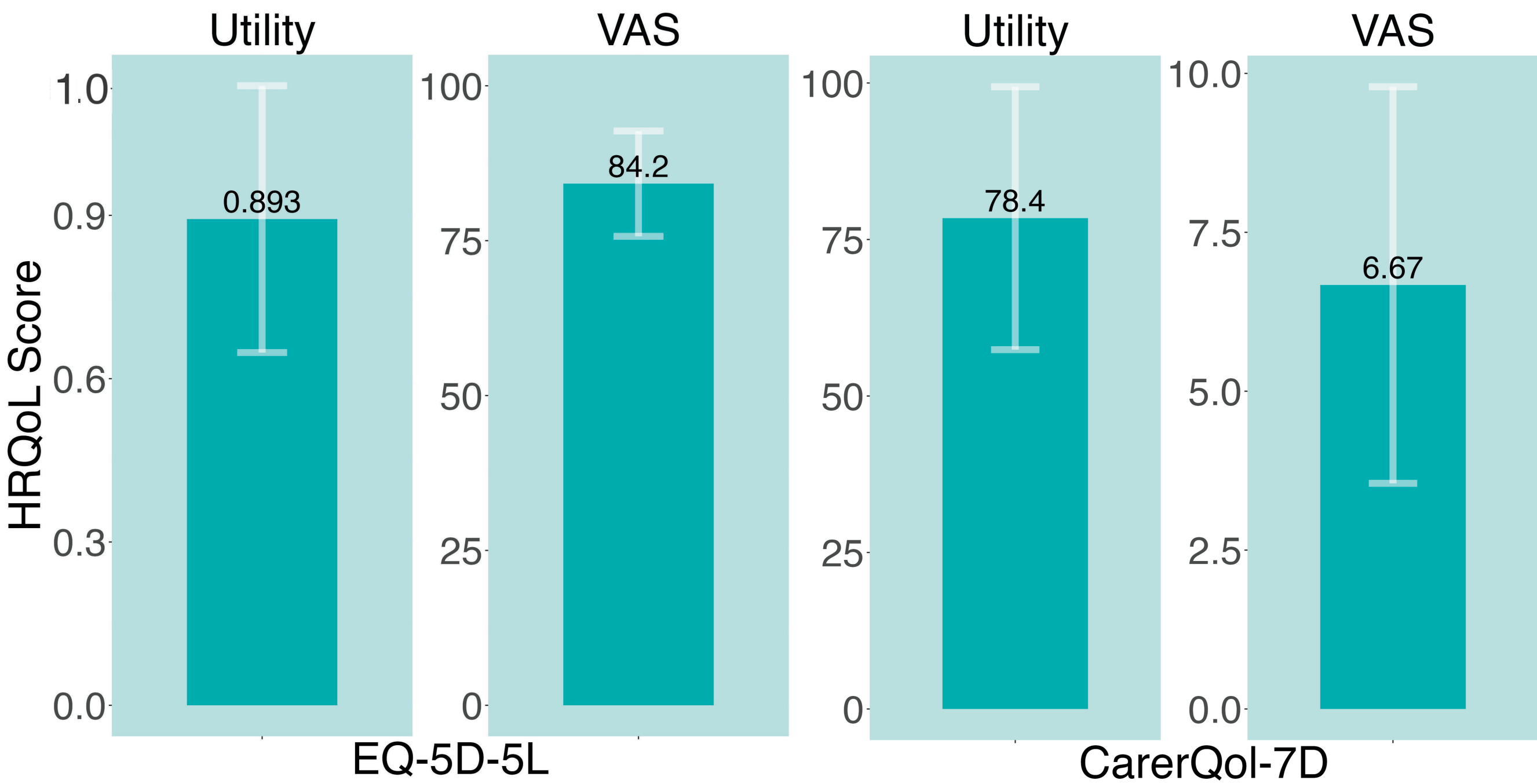


Figure 1. Mean EQ-5D-5L and CareQoL-7D utility value and VAS scores

- **WPAI:** Most carers were in employment (n=7, 78%). Mean absenteeism was 14%, however, this value was driven by a single carer who was currently unable to work.
- Of those in work, mean presenteeism was 15%.
- Mean work productivity loss was also 15% and mean activity impairment was 36.7%, see **Figure 2**.

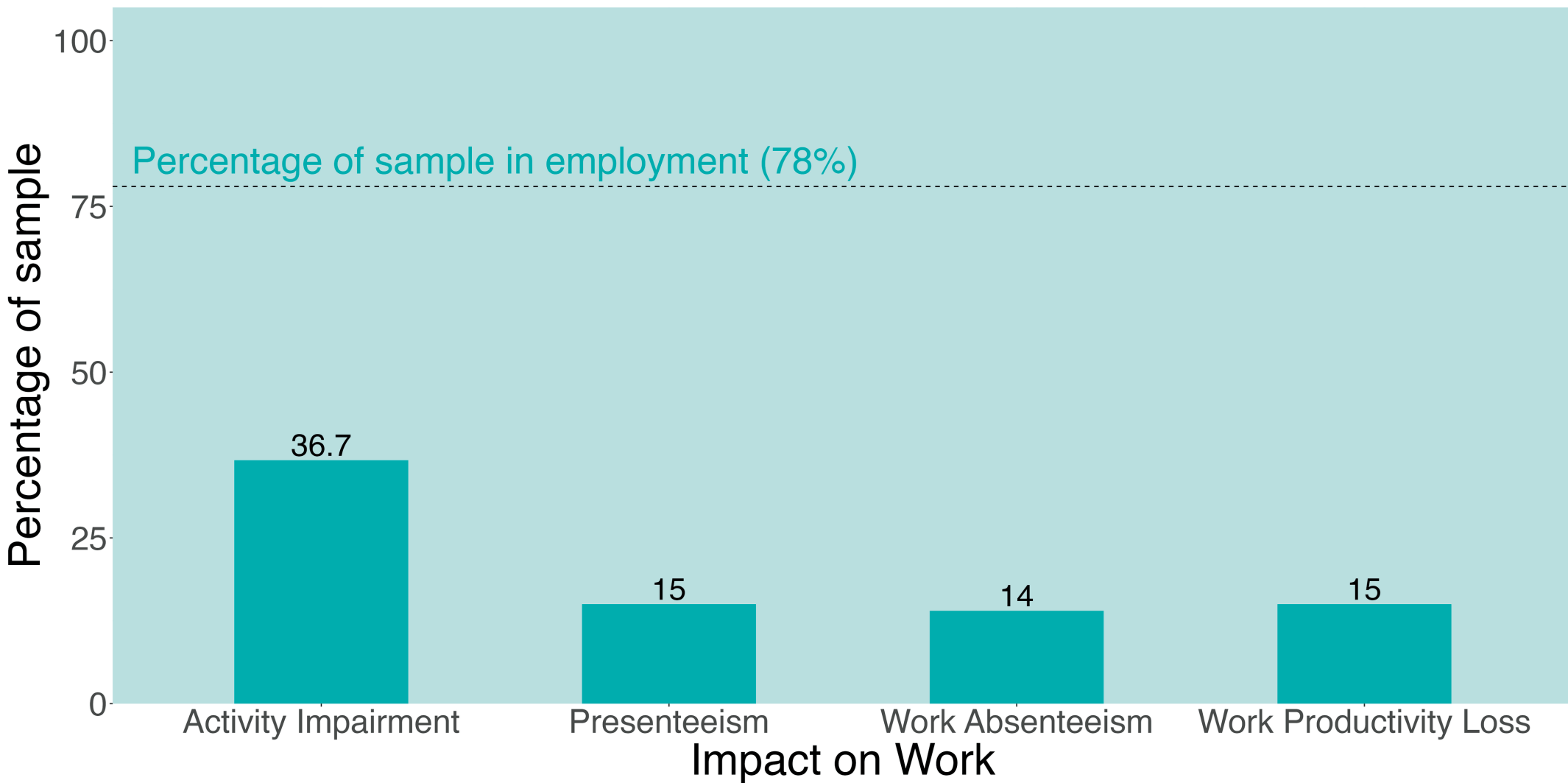


Figure 2. Mean WPAI domain scores

QUALITATIVE RESULTS

- Qualitative findings (N=9) revealed substantial burden for many carers and family members.
- The most prominent impacts were emotional (e.g., guilt, devastation), including many participants who described the specific emotional impact of LHON being a maternally inherited disease (n=8/9 were female).
- Impacts to daily life, social life and relationships, work and career, finances and the wider family were also described.
- A summary of carer and family impacts is presented in **Figure 3**.

Carers reported feelings of devastation at the time of diagnosis. Mothers discussed immense feelings of guilt for passing on a gene that caused their child's vision loss. Worry about the future was a prominent theme across interviews. The profound emotional impacts of caring for someone with LHON had knock on effects on other areas of life.

Carers discussed how their daily routine and activities had changed to accommodate their care tasks. Several carers reported that they prioritised the individual with LHON's daily activities over their own and had limited time to participate in leisure activities and hobbies.

"Watching your firstborn child's every plan, every hope, every dream change overnight, that's absolutely devastating" CG05, parent

Due to the genetic nature of the condition, carers reported that family members worried about themselves or their children developing symptoms in future to the extent that some sought professional help to cope with this anxiety, while others decided against starting a family or having more children.

"Mum, I've got a disease that I've passed onto my child, it could cause him to go blind in the future", she said, 'and I was about to have another child', she said, 'and now I can't" CG01, parent

Financial impacts included the cost of travel and accommodation for appointments and self-funded treatment. Carers who had a partner with LHON who did not work, supported them financial and reported limited money for themselves or luxuries such as holidays.

"Financially, it's quite hard because I get all the mortgage taken out of my account and I buy all the food... I'd love just to be able to go out and do a massive shopping trip to myself but the end of the month, normally we don't have that much left" CG07, partner/spouse

"I have to spend more time at home than perhaps the average person would do, they'd get more involved in outdoor activities, clubs, societies, all that sort of thing, which is not really viable for me" CG04, partner/spouse

Some carers described the limited time they had to socialise, while others reported intentionally isolating themselves to avoid discussing LHON with friends. One carer had even cut ties with friends due to a lack of support and understanding. A few carers reported that the physical and emotional demands of providing could cause a strain on their relationship with their partner.

"There's a lot of friends I don't really want to see because I don't want to sit and talk about it" CG01, parent

Most carers had taken time off work around the time of diagnosis to provide support. One participant had been signed off work due to the emotional impacts of the diagnosis, while others had reduced their hours and work trips to cope with the logistical and emotional demands of providing care.

"I've needed to be part time really, to have the time and emotional energy to deal with things that suddenly crop up" CG08, parent

Figure 3. Map of carer impacts from qualitative analysis

DISCUSSION AND CONCLUSIONS

- The qualitative findings suggest a substantial burden for many carers and family members with impacts reported across numerous aspects of daily life.
- However, the burden described in the qualitative data was incongruent with the quantitative measures.
- Questionnaire results suggest a relatively small burden to carers with minimal impacts on HRQoL, work and regular activities reported. For example, the mean EQ-5D utility was 0.89, compared with a UK population average of 0.85 in adults aged 45-54 [8].
- This demonstrates the value of conducting mixed-methods research to understand the impact of disease and the importance of selecting measures which capture population-relevant concepts.
- A potential limitation of the current study is the sample size was small (N=9) and included three participants which were a carer or family member of the same individual with LHON meaning their experiences may have been disproportionately represented.
- **This research demonstrates the substantial burden associated with LHON and potential spillover effects on carers and family members.**

REFERENCES

- 1.Shemesh et al. StatPearls Publishing 2022:1-3.
2. Mackey DA, et al. Am J Hum Genet 1996;59:481-5.
3. Harding AE, et al. Am J Hum Genet 1995;57:77-86.
4. Man PYW, et al. J Med Genet 2002;39:162-169.
- 5.Chen, BS et al. J Neuro-Ophthalmol 2022;00:1-7.
- 6.Hernández-Alava, M et al. Health Econ 2022;31:923-939.
7. Hoefman, RJ et al. Pharmacoeconomics 2017;35:469-478.
8. Szende, A et al. Springer 2014.

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