



Retinitis Pigmentosa: Disease Encumbrance in the Eurozone

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Abstract

Introduction: Retinitis Pigmentosa (RP) is the most common hereditary progressive retinal degenerations and is first cause of irreversible blindness in developed countries among persons under 70 years of age. It is characterised by progressive degeneration of the retinal photoreceptors. The objective of this review was to determine the burden of RP in the Eurozone.

Methods: A targeted literature search was conducted and published literature was reviewed for studies depicting the burden of RP, expressed as diminished vision, blindness, and quality of life. We used combinations of the terms like; "burden", "impacts", "effects" and "review" to search for eligible studies in Medline, EMBASE, and Web of Science databases. Grey literature searches were conducted in Google Scholar and SIGLE (System for Information on Grey Literature in Europe). Additionally, reference lists of reviewed studies and articles were screened for titles that included key terms.

Results: Epidemiological studies conducted in the United States, Switzerland, Slovenia, Denmark, and England show an estimated overall prevalence of RP approximately 1/4000 in these countries. Five to seven per cent of newly diagnosed blindness in the western countries is attributable to RP and it represents one of the most common causes of blindness or severe diminished vision in people within 20 to 60 years old. The estimated population affected by RP in Europe is 167000. In Denmark the lifetime risk of developing RP is 1/3000, and in Sweden it is estimated to be 1/2000. The prevalence for 2013 based on the population according to the World Health Organization for different countries is extrapolated.

Conclusion: RP is associated with both significant economic impact and reduction in quality of life. It raises serious social challenges for both patients and their families, interfering with day-to-day life.

Keywords

Retinitis pigmentosa, Diminished vision, Blindness, Quality of life, Burden, Impact, Effects and review

Introduction

Retinitis Pigmentosa (RP) is an inherited retinal dystrophy of the posterior segment of the eye that causes severe vision impairment. It is characterised by progressive degeneration of the retinal photoreceptors, deposition of retinal pigments predominantly in the peripheral retina and sparing of the central retina [1-3]. Retinitis Pigmentosa is a bilateral inherited condition that involves both eyes. It usually starts later in life and progresses to blindness. RP is the foremost source of irreversible

blindness in developed countries among individuals under 70 years of age. Symptoms characteristically begin in the early teenage years, and severe visual impairment occurs by ages 40 to 50 years. The symptoms include night blindness followed by diminishing visual fields, leading to tunnel vision and eventually legal blindness or, in many cases, complete blindness. In most of the cases of RP, there is a primary degeneration of the photoreceptor rods, with secondary degeneration of cones. Ageing population is constantly ascending in the developed countries and diminished vision and blindness are primarily problematic [4]. There are no real treatment options. In the developed countries the health-related quality of life in patients with severe visual impairment is found to be similar or even less and with higher emotional disturbance as compared with patients having chronic health issues like stroke and tumours [5]. Management of the individuals having moving difficulties and loss of reading due to blindness necessitates psychological help.

RP has a worldwide prevalence of 1/3000 persons to 1/7000 persons [6]. Prevalence of non-syndromic RP is approximately 1/4,000 [3]. It is estimated to affect approximately 1.5 million people worldwide [7]. There is significant cost burden as a result of blindness and diminished vision impact not only to the patient but also to the family, caregivers and the community [8]. As demands on healthcare continue to increase in the developed countries, economic evaluations of the diseases and interventions have also become progressively imperative [3]. There are direct costs, indirect costs and intangible costs associated with visual impairment.

Keeping the potential constraints like limited healthcare resources, lack of clear understanding about the economic impact of RP and various costs associated with it we, conducted a comprehensive literature review, amalgamating entire possible data available. The objective of this review was to determine the burden of RP in the Eurozone.

Methodology

A targeted literature search was conducted and published literature was reviewed for studies depicting the burden of RP, expressed in diminished vision, blindness, and quality of life. We used combinations of the terms like; "burden", "impacts", "low vision", "visual impairment", "visually impaired", "blindness", "and blind", "visual loss", "costs, costs of illness", "effects" and "review" to search for eligible studies in Medline, EMBASE, and Web of Science databases. Grey literature searches were conducted in Google Scholar and SIGLE (System for Information on Grey Literature in Europe). Additionally, reference lists of reviewed studies and

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Table 1: Extrapolated Prevalence of Retinitis Pigmentosa in the Eurozone 2013

Country	2013 population	Extrapolated Prevalence
United Kingdom	63,136,000	23360
Germany	82,727,000	30609
France	64,291,000	23788
Italy	60,990,000	22566
Spain	46,927,000	17363
Netherland	16,759,000	6201
Switzerland	8,078,000	2989

articles were screened for titles that included key terms. As we were concerned in the burden of RP in high-income countries specifically The Netherlands, The United Kingdom, Germany, France, Italy, Spain and Switzerland, we excluded economic studies conducted in the developing countries. Provision of health services and disease management options vary immensely between countries, making an assessment of cost categories unfeasible.

Results

Five to seven per cent of newly diagnosed blindness in western countries is attributable to RP and it represents one of the most common causes of blindness or severe low-vision in people from 20 to 60 years old [9]. The estimated French population affected by RP is approximately 24000 and in the whole Europe total numbers of people affected are 167000 [10]. Extrapolated prevalence of RP in seven EU countries based on their year 2013 populations is presented in the table 1 [11,12].

Epidemiological studies conducted in the Switzerland, Slovenia, Denmark and England show an overall estimated prevalence of RP approximately 1/4000 [13-15]. However, the prevalence in each country varies. In Sweden the estimated RP population was found to be 1/2000. Furthermore, in Switzerland, the prevalence of RP has been reported to be as low as 1/7000 [11]. The statistics used for prevalence of RP as depicted in table 1 are typically based on US, UK, Canadian or Australian prevalence statistics that is further extrapolated using only the population of the respective country.

Discussion

In spite of the lack of treatments, regular general eye check-ups are imperative for RP patients because people with RP are still at risk for other eye problems which may be treatable. Patients with RP tend to develop cataracts at an earlier age than the individuals without RP. There is higher risk of injury, [16-19] difficulties with timely access to health care, [19] greater dependency and increased rates of admission to institutional amenities, [18] and reduced mental well-being [20]. There is increase in the risk of cardiovascular, metabolic, and psychiatric disorders [21]. RP patients suffer from depression [22] and there is also productivity loss [23]. RP is the most common of the retinal degenerations. It is the leading cause of inherited blindness in the developed world. Rare diseases, including those of genetic origin such as RPs, are life-threatening or chronically debilitating diseases. In a developed country with ageing population visual loss has substantial impacts on the quality of life. The projected prevalence depicted in this review confirm the need to develop and consume treatments and technologies to prevent visual loss. This can be accomplished with cost effective interventions. To enhance the quality of life and reduce the impact of visual loss there is a need for rehabilitation services for patients with RP. Further research is needed to develop models of effective services that can provide for greater numbers of people with diminished vision. Explicit initiatives and health policies need to be established to address this issue. In conclusion, the overall findings of this review raise alarms. Prevalence of RP and its burden in the Eurozone is high. The enduring burden of RP and associated vision loss points to an urgent need for further research as it raises serious social challenges for both patients and their families, interfering with day-to-day life. In conclusion, RP cause a considerable burden on affected persons, their caregivers and society at large, which increases with the degree of visual impairment. Public and private investments to research new treatments will benefit both patients and the economy. According to the European Federation of Pharmaceutical Industries and Associations

(EFPIA), in an aging society and with the continuing economic crisis, European citizens are optimistic with regards to innovative treatments which will enable them to live longer, more active and healthier lives [24].

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