

# Daily life, experience and needs of persons suffering from homozygous familial hypercholesterolaemia: Insights from a patient survey

Eric Bruckert <sup>a,\*</sup>, Samir Saheb <sup>b</sup>, Juliette Roth Bonté <sup>c</sup>, Carole Coudray-Omnès <sup>c</sup>

<sup>a</sup> *Department of Endocrinology and Cardiovascular Disease Prevention, Hôpital Pitié-Salpêtrière, Paris, France*

<sup>b</sup> *Haemobiotherapy Clinical Centre, Hôpital Pitié-Salpêtrière, Paris, France*

<sup>c</sup> *Cemka Eval, Bourg-La-Reine, France*

## Abstract

Homozygous familial hypercholesterolaemia (HoFH) is a rare and severe hereditary lipid disorder that is typically associated with high serum levels of low-density lipoprotein cholesterol (LDL-C). Excessive exposure to high levels of LDL-C puts affected individuals at very high risk of premature onset coronary heart disease, and this considerably limits life expectancy. Although the clinical features and treatment of HoFH have been extensively researched, societal and socio-psychological impacts of the disease have not been reported to date. The current study was conducted to investigate the burden of disease and treatment from the patient's perspective by means of semi-structured interviews with 24 HoFH patients. The findings of the survey indicate that HoFH represents a considerable burden for patients, not only due to physical signs and limitations caused by the disease but also a number of psychosocial factors, treatment-related issues and impact on their education and employment situation.

© 2014 Elsevier Ireland Ltd. All rights reserved.

**Keywords:** Homozygous familial hypercholesterolaemia; Patient survey; Burden of disease; Psychosocial factors

## 1. Introduction

Homozygous familial hypercholesterolaemia (HoFH) is a rare and severe hereditary lipid disorder that is typically associated with high serum levels of low-density lipoprotein cholesterol (LDL-C). However, levels of serum LDL-C show considerable variability, depending on the genetic defect and whether or not the patient is receiving lipid-lowering treatment. Reports from the literature suggest that LDL-C levels in HoFH can reach around 450–1000 mg/dL, and that treatment can reduce this to between 220 and 600 mg/dL [1–3].

The impact of HoFH on morbidity and mortality is well documented. Excessive exposure to high levels of LDL-C puts affected individuals at very high risk of premature onset coronary heart disease, and this considerably limits life expectancy [4–6].

The clinical features and treatment of HoFH have been extensively researched. A review of literature indexed on PubMed performed in March 2013 identified a total of 1185 publications on HoFH [7]. These were mainly related to the diagnostic criteria, aetiology, clinical presentation, management, treatment options, epidemiology and economic burden. Despite data having been collected on a wide variety of HoFH related topics, societal and socio-psychological impacts of the disease have not been reported to date. Moreover, the literature search did not identify any studies detailing the experience of persons affected by HoFH (Health-Related Quality of Life Patients Reported Outcomes). To address this shortfall, the burden

\* Corresponding author. Service d'Endocrinologie Métabolisme, Groupe Hospitalier Pitié-Salpêtrière, Assistance Publique – Hôpitaux de Paris, 47-83 Boulevard de l'Hôpital, 75651 Paris Cedex 13, France. Tel.: +33 1 42 17 57 85; fax: +33 1 42 17 78 65.

E-mail address: [eric.bruckert@psl.aphp.fr](mailto:eric.bruckert@psl.aphp.fr) (E. Bruckert).

of HoFH and its treatment was investigated from the patient's perspective in a research study conducted at the Haemobiotherapy Clinical Centre, Hôpital Pitié-Salpêtrière, which receives the majority of HoFH cases in France.

## 2. Methods

### 2.1. Development of questionnaire and recruitment and selection of interviewees

An interview questionnaire was developed to understand what daily life with HoFH is like, focussing on the personal, familial, social and psychological impact of the disease.

The investigating physicians identified persons meeting the inclusion criteria and invited them to participate in the survey. The study included adult HoFH patients aged over 18 years who had given written informed consent for data to be collected and used anonymously within the context of this survey.

### 2.2. Interview methodology and analysis

Semi-structured interviews, which lasted an average of 45 min, were conducted within the Haemobiotherapy Unit of Hôpital Pitié-Salpêtrière in March and April 2013. Each interview was transcribed and an analysis of the quantitative questions of the questionnaire was performed. The findings presented in this paper are illustrated with the participants' own comments, which have been quoted verbatim (following translation from French into English).

## 3. Results

### 3.1. Patient profiles

Twenty-four interviews were conducted with 13 women and 11 men aged 18–54 years who had attended for apheresis at the centre for several years. Four patient profiles were identified from the 24 interviews.

1. *Patients who considered themselves as “not ill”* ( $n = 2$ ). These were patients diagnosed with HoFH who were immigrants to France, arriving in the country at around 20 years of age. The treatments that they received since arriving in France (medicines, apheresis and cardiac surgery) have had an apparently curative effect, to the extent that these patients reported feeling “normal ...”.
2. *Patients who considered themselves as “normal”* ( $n = 11$ ). These patients reported the disease as present only on the day of apheresis treatment, and at other times they do not think about it. From their point of view, they live normally.

3. *Patients who considered themselves “sick intermittently”* ( $n = 5$ ). For these patients the disease was fairly present in their everyday lives, either from a physical or psychological perspective. In spite of the disease's intermittent presence, however, they felt they lived normally.

4. *Patients who considered themselves “sick”* ( $n = 6$ ). For these patients the disease was slowly getting the upper hand in their life. Unlike patients in the other three groups, they lived daily with a disease that weighed heavily on them.

### 3.2. History of cardiovascular disease: summary of patient-reported events

Although the survey was not designed to assess the cardiovascular disease history of the patients, during the interviews, more than half of the cohort (13/24; 54%) reported severe cardiovascular events.

Four patients had undergone carotid artery angioplasty with stenting (the number of stent placement procedures varied from 2 to 7 per patient). Three patients reported bypass surgery and in two cases this was associated with an aortic valve replacement. One patient had undergone aortic valve replacement twice and, in total, five patients had had one or two valves replaced.

Other cardiac events reported included (one case of each): cardiac arrest, angioplasty of the left coronary artery, heart implant and cardiac surgery (specific procedure not described). For two patients, the cardiovascular events (cardiac arrest and double bypass procedure, respectively) occurred during childhood before the age of 10 years.

These data clearly illustrate the cardiovascular outcomes directly associated with HoFH.

### 3.3. Distance travelled to treatment centre and time spent on patient care

The mean distance patients travelled to the apheresis centre was 184 km (range 4–500 km; standard deviation 193 km); median distance was 90 km. Twelve patients travelled up to 100 km, four patients travelled between 100 and 200 km and eight patients between 300 and 500 km. Patients used a variety of different means of transport to reach the apheresis centre (Table 1). In France, the cost of apheresis treatment and patients' transport costs are fully covered by the social security system and most of the patients in this study ( $n = 18$ ) reported that their travel expenses were fully covered by social security or state medical assistance.

On average patients spent 5 h per week (median 4.5 h) on their care, which included everything that they had to do for their daily care, travel time and treatment time. Nineteen patients spent between 2 and 4.5 h on their care and five patients spent between 6 and 12 h. The actual duration of treatment varied from 2 to 4 h depending on technical

Download English Version:

<https://daneshyari.com/en/article/2895435>

Download Persian Version:

<https://daneshyari.com/article/2895435>

[Daneshyari.com](https://daneshyari.com)