Williams KR et al., J Genet Syndr Gene Ther 2018, Volume 9
DOI: 10.4172/2157-7412-C1-018

Results of the investigation of findings and observations captured in burden of illness survey in FCS patients (in-focus) study: European respondents

Williams KR[1], Davidson M[1], Stevenson M[2], Hsieh A[2], Jones R[2], Issa B[3], Romeo S[4]
[1]Akcea Therapeutics™, Inc., USA;
[2]Department of Medicine, University of Chicago, USA
[3]Department of Endocrinology, University Hospital of South Manchester, UK
[4]University of Gothenburg, Sahlgrenska Academy & Sahlgrenska University Hospital, Department of Cardiology, Sweden

Introduction: Familial Chylomicronemia Syndrome (FCS), is a rare autosomal recessive disease characterized by extremely high serum triglycerides (TGs), carried in chylomicrons, which predisposes the patient to recurrent episodes of abdominal pain and risk of acute pancreatitis (AP). The physical, emotional, psychosocial and cognitive consequences of living with FCS are poorly understood and not documented in the literature.

Methods: The In-FOCUS web-based patient survey was undertaken to quantify the burden of illness and quality of life from the patient's perspective.

Results: 14 adult FCS patients from 5 European countries completed the survey. The majority of patients (64.3%) reported that their disease adversely affected their life over the past 12 months; with their stress/anxiety level (64.3%), ability to socialize (57.1%), ability to travel for work or leisure (57.1%), their mental ability (53.8%), quality of sleep (50.0%), and their feeling of self-worth (50.0%) all impacted. Over the past 12 months, 42.9% of patients had to take an average of 10.8 days off work because of problems related to FCS. 58.3% of patients felt their disease had influenced their decision on whether to have children, or how many children to have. 57.1% of patients reported feeling a burden to those around them because of their FCS.

Conclusions: FCS imparts a marked burden to the patient which extends beyond the recognized physical symptoms.

Recent Publications:

1. Davidson, M., et al. The burden of familial chylomicronemia syndrome: interim results from the IN-FOCUS study. *Expert Review of Cardiovascular Therapy*. Mar 2017.
2. Gelrud, A., et al. The burden of familia chylomicronemia syndrome form the patients' perspective. *Expert Review of Cardiovascular Therapy*. Sept 2017.
3. Ahmad, Z., et al. Building a better understanding of the burden of disease in familial chylomicronemia syndrome. *Expert Review of Clinical Pharmacology*. Nov 2016.

Biography

Dr.Karren Williams has experience in the pharmaceutical industry with previous leadership roles at Akcea Therapeutics. She was intricately involved in the design and refinement of the study survey as well as being largely responsible for study approval and access in the United States and Canada. Previously, she was also heavily involved in the largest global patient voice quality of life study in patients with FCS conducted by Akcea Therapeutics (IN-FOCUS).

bhubbard@akceatx.com