

Examining the high disease burden and impact on quality of life in familial chylomicronemia syndrome

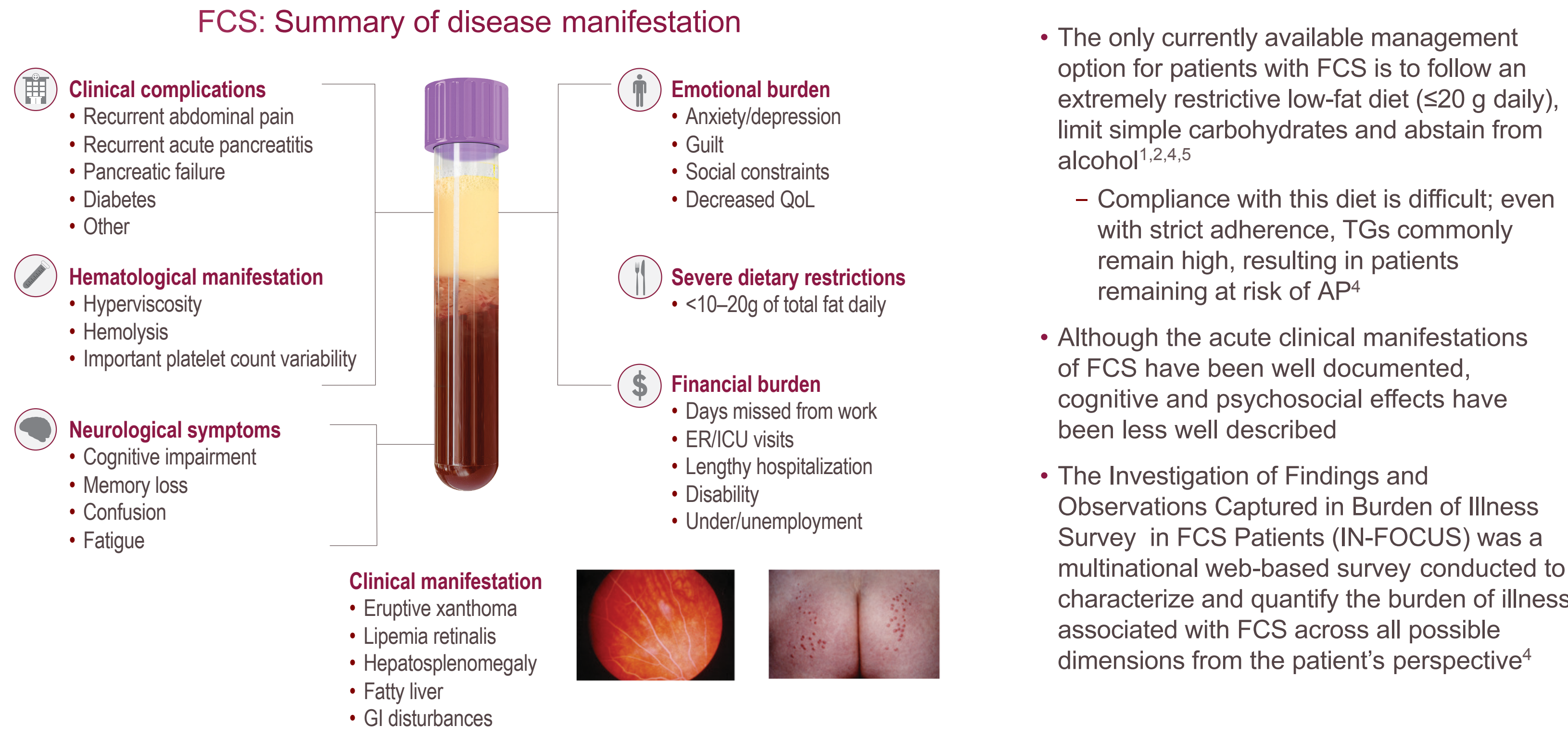
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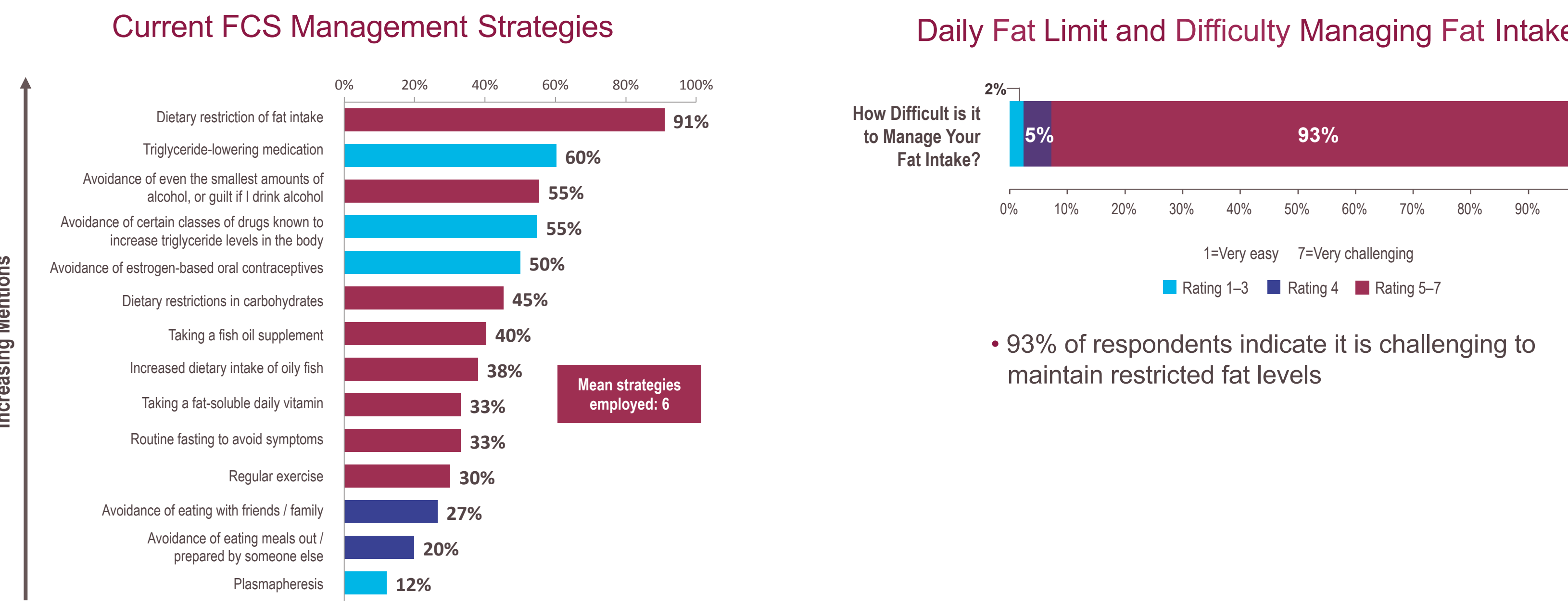


Introduction

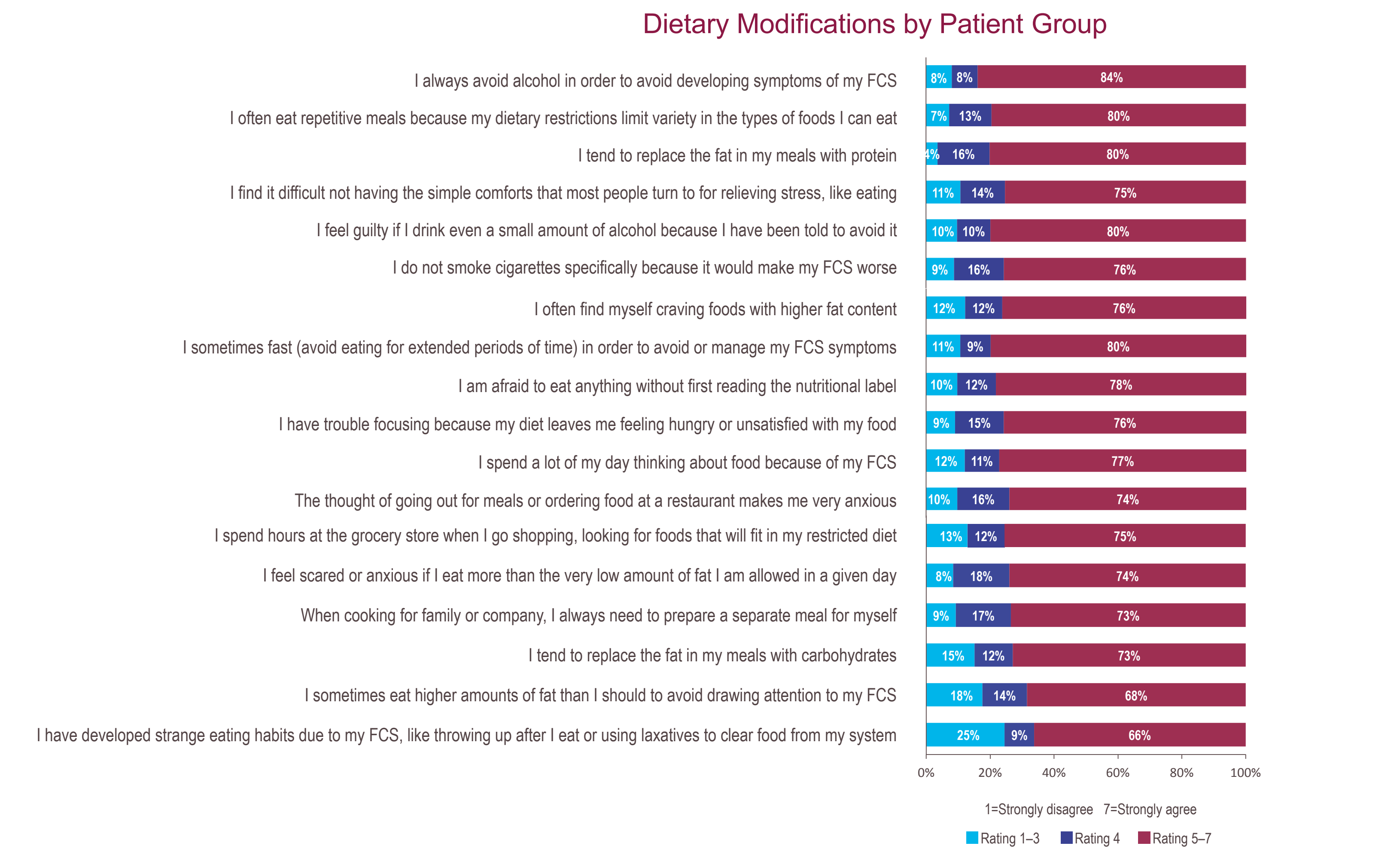
- Familial chylomicronemia syndrome (FCS) is a rare genetic disorder characterized by severe chylomicronemia, leading to high serum triglycerides (TGs) and consequent risk of recurrent and acute, potentially fatal, pancreatitis²
- FCS is caused by a deficiency in the production or functionality of lipoprotein lipase (LPL), the enzyme that breaks down plasma TGs¹
- Patients with FCS typically present with multiple symptoms as a result of severely high TG levels, including intense abdominal pain, nausea and vomiting, and fatigue¹⁻⁴



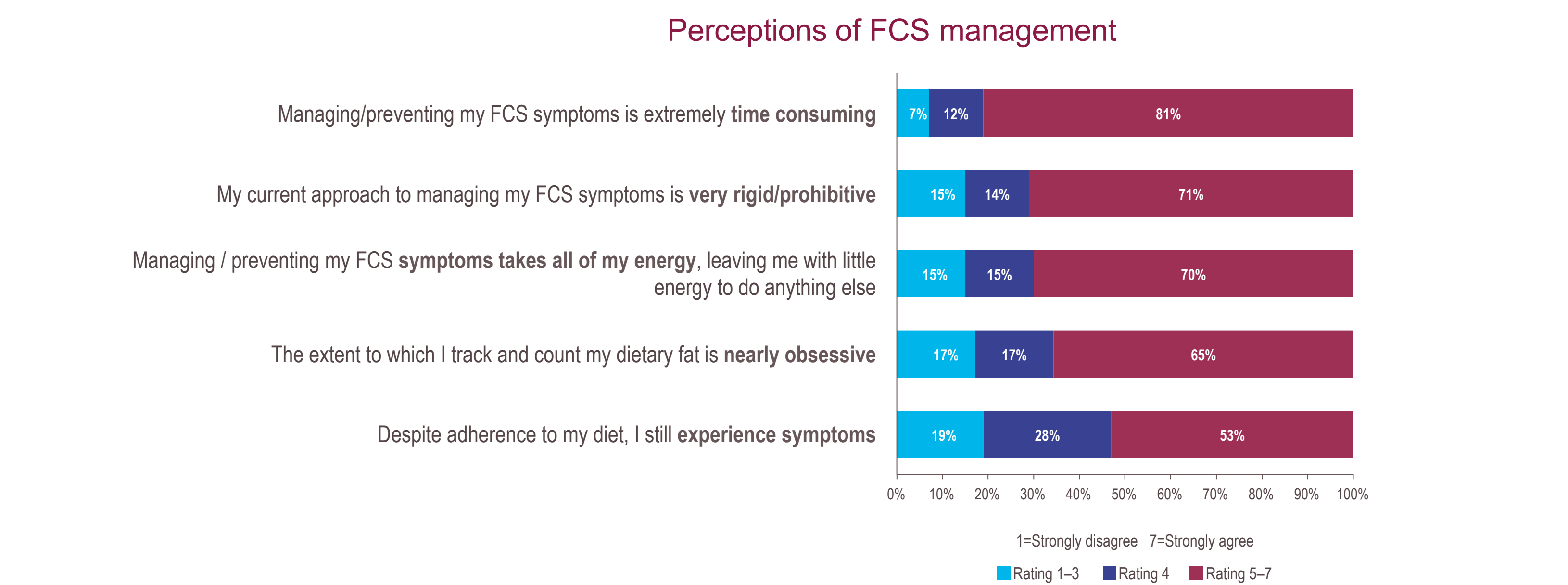
Results



- FCS patients not only seek to restrict fat from their diets, but also employ other methods to attempt to reduce TGs and potential symptoms, including pancreatitis
- On average, patients reported using six different approaches to reduce plasma TGs



- Dietary adherence is not only restrictive, but can also affect patients' mental/emotional health
- The majority of respondents strongly agree that they make extreme dietary restrictions as a means to control their FCS symptoms and strongly agree that they experienced food cravings (76%) and have trouble focusing due to restricted diet (76%)
- The majority of respondents felt scared/anxious if they exceeded the permitted daily level of fat intake (74%), and spend considerable time choosing and preparing meals due to FCS (73%)



- 70%, 81%, and 71% of respondents find managing symptoms extremely energy draining, time consuming, and rigid/prohibitive respectively
- Despite adherence to diet, 53% of respondents still experienced symptoms associated with FCS

Conclusions

Largest Burden of Illness study in patients with FCS (N = 166) to date, filling a major gap in the literature

Patients face many comorbidities associated with FCS, the most serious of which is acute pancreatitis

- 40% of patients reported experiencing an event of pancreatitis
- Average of 13 events over their life so far

References

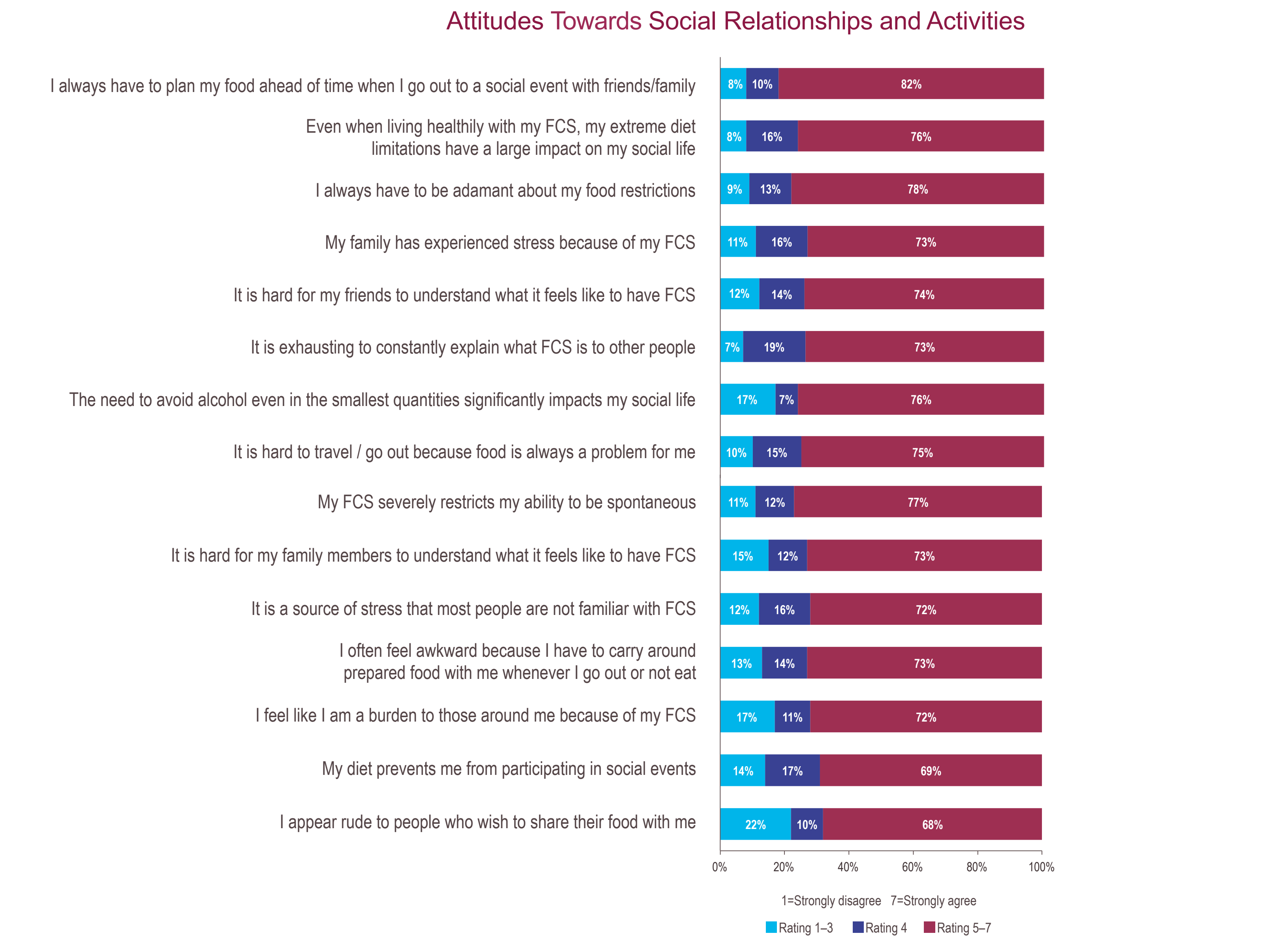
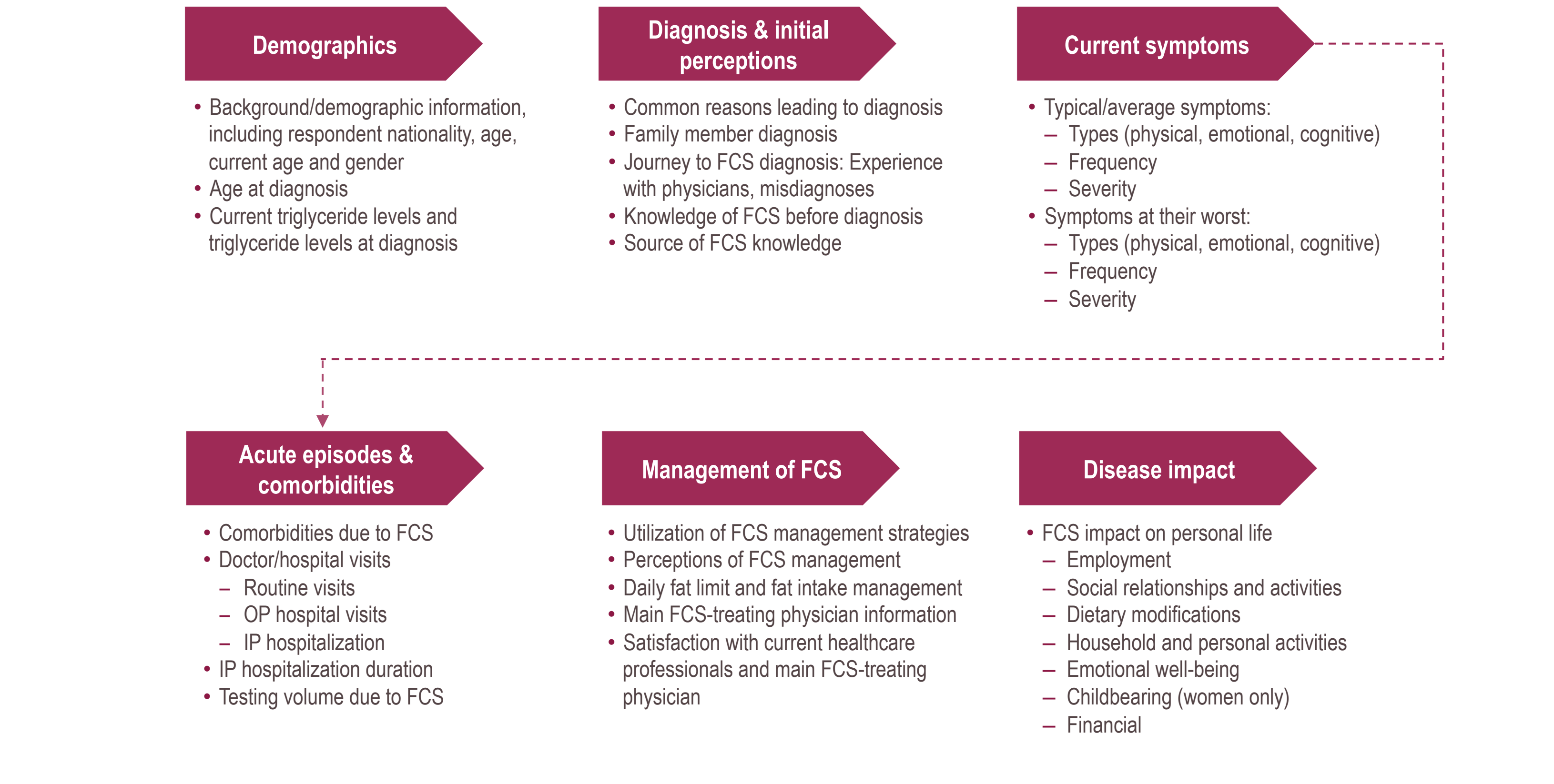
- Gaudet D et al. *N Engl J Med* 2014;371:2200–2206.
- Brahm AJ and Hegde RA. *Nat Rev Endocrinol* 2015;11:352–62.
- Symersky T, van Hoon B, Masclee AAM. *JOP* 2006;7:447–53.
- Davidson M et al. *Expert Rev Cardiovasc Ther* 2017;15:415–423.
- Valdivielso P, Ramirez-Bueno A, Ewald N. *Eur J int Med* 2014;25:689–694.

Disclosures

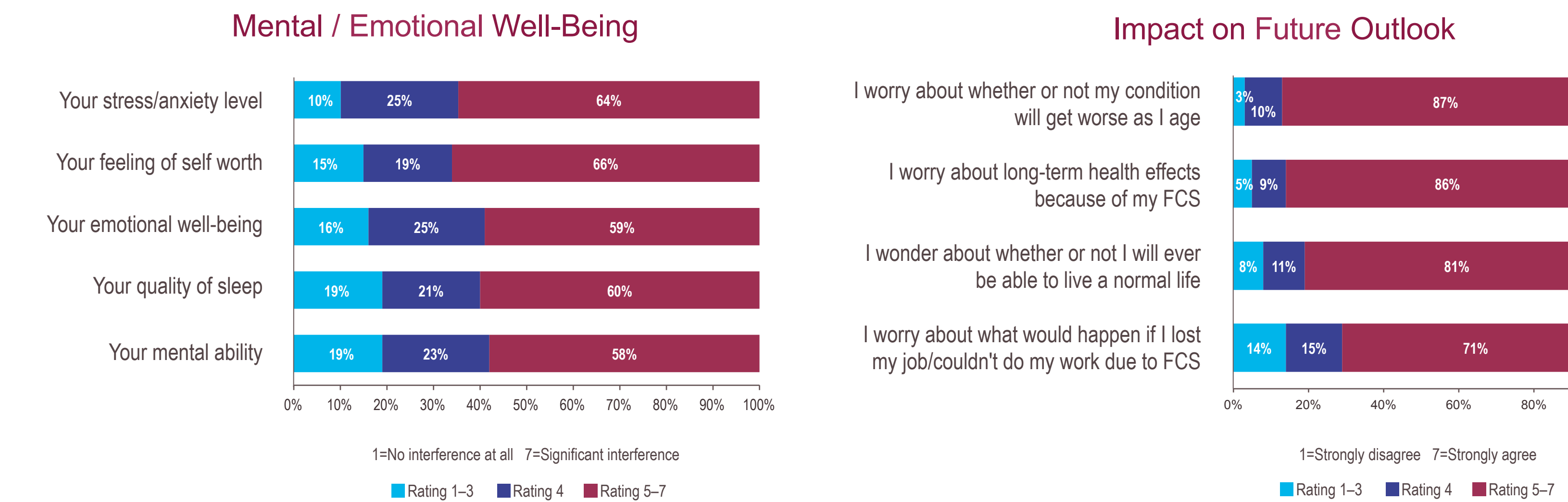
- This study was funded by Akcea Therapeutics, Inc.
- Michael Davidson is a scientific advisory board member of Abbott, Amgen, AstraZeneca, Merck and Sanofi, Regeneron.
- Michael Stevenson, Andrew Hsieh, and Karren R. Williams are employees of Akcea Therapeutics, Inc.
- Zahid Ahmad has received research support from the National Institutes of Health, Regeneron and the FH Foundation, and has received honorarium from Amgen and Sanofi, Regeneron.
- Jeanine Roeters van Lennep has received grants from the Dutch Heart Foundation, serves on the Governance advisory board of Amryt and has received honorarium from Akcea Therapeutics, Inc.

Methods

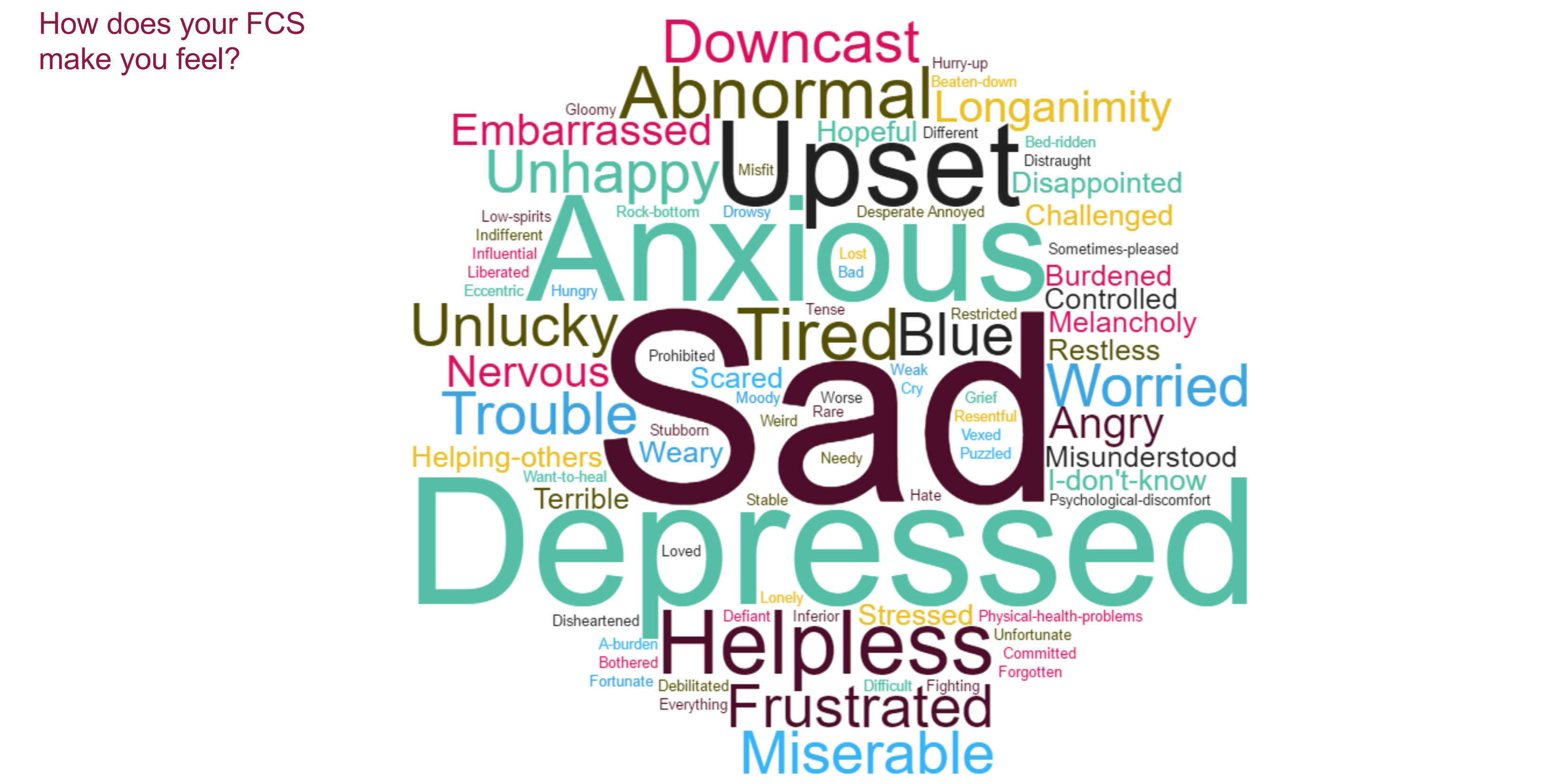
- IN-FOCUS was a self-reported, online, anonymous quantitative research survey (approx. 45 mins), conducted with patients diagnosed with FCS (n = 166)
- All research materials were approved by relevant country-level ethics committees



- Respondents felt socially restricted because of their FCS; most reported that FCS affects multiple aspects of their social life and relationships



- FCS significantly interferes with mental and emotional well-being across multiple domains
- A vast majority of respondents reported being concerned about the potential long-term impact of FCS on their health as well as other aspects of their life, including work



The burden of FCS goes far beyond physical symptoms & impacts patients' mental & emotional well-being & even professional endeavors

- Only 23% of patients reported full time employment, with 65% of unemployed patients reporting it was largely due to FCS
- FCS negatively affects patients stress/anxiety levels, mental ability, and feeling of self worth

Management strategies are incredibly challenging and time consuming to adhere to

- 53% of patients reported still experiencing symptoms even with rigorous adherence
- Diet is a frequent source of anxiety, worry and fear for patients with FCS and make social activities extremely challenging

Acknowledgments

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