The burden of familial chylomicronemia syndrome (FCS) in Brazilian patients:
A pilot qualitative study

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BACKGROUND

Familial chylomicronemia syndrome (FCS) is a very rare, genetic disease that prevents the body from breaking down fats (triglycerides). It is estimated to affect 1 individual per one million but the prevalence can be higher in some areas. Males and females are equally affected.[1]

Symptoms of FCS include eruptive xanthomas (raised yellow papules surrounded by an erythematous halo), lipaemia retinalis (lipaemic retinal vessels observed on fundoscopy), hepatosplenomegaly or splenomegaly, nausea, vomiting, failure to thrive and recurrent abdominal pain.[2,3] FCS patients may also present acute pancreatitis episodes, which are potentially life-threatening, and may develop chronic pancreatitis.[2] Patients may also have emotional and psychosocial effects, including anxiety, social withdrawal, depression and brain fog.[2] Symptoms onset can be as early as infancy, but some patients may not present symptoms until early adulthood.[3].

FCS in Brazil is often misdiagnosed and characterized by low clinical awareness that negatively affects the patient journey and experience, often resulting in inadequate care.[4]

Additional research to assess the FCS patient burden in Brazil **could provide insights to refine healthcare services and improve outcomes.**

This is the 1st **qualitative study in Brazil** aimed to understand the burden of FCS in patients.

METHODS

In-depth face-to-face semi-structured interviews were conducted in June 2022. The Brazilian interviews aimed to capture patients' journeys, and identify their needs, motivations and perspectives about FCS.

8 patients:
1 male / 7 females
Age range:
40-55 years old

Transcripts were analyzed thematically in an iterative process.

RESULTS - Patient Burden

Patients commonly reported the following **three life impact themes:**

1

Psychological Burden (Frequency 8/8)

Patients perceived that the greatest impact of FCS was on their emotional well-being, **with all 8 patients reporting depression**

2

Medical Problems (Frequency 8/8)

Medical / physical problems were described by patients and caregivers. **All 8 patients experienced acute pancreatitis and reported severe abdominal pain**

3

Interpersonal Problems

Diet was reported as a frequent source of anxiety by patients, preventing them from participating in social activities and causing interpersonal problems, including constraints in professional, personal, and financial life, stigmatization and a lack of understanding by others

Other less frequent themes mentioned by patients:

Pregnancy Burden (Frequency 2/8)

Two patients reported that **living with FCS impacted their decision to have children,** perhaps this is linked to the risk of FCS during pregnancy

Socioeconomic Burden

Socioeconomic factors were considered crucial to their psychological and interpersonal problems and adherence to a low-fat diet

DISCUSSION - Socioeconomic Status

When discussing experiences and perceptions of FCS patients in Brazil, and particularly the quality of care that is provided to them, **it is fundamental to address some social, economic and cultural issues of the population** - mainly of the underserved patients.

Difficulty accessing basic public services

- Education, health, basic sanitation
- Few leisure options and free access to culture
- More dependent on the public services and federal government programs to ensure access to basics services

Low levels of education

- Most of the interviewed patients are illiterate or have only completed secondary education, which is often incomplete.

Unemployment, underemployment, informality

- Jobs with high physical demands, with greater exposure to health risks
- Underpaid jobs, making it difficult to adhere to a proper high-cost diet

Housing issues

- Precarious, small, unfinished homes without ventilation
- Lack basic items like a computer or internet

Urban violence

- They live in peripheral neighborhoods where the incidence of crimes is higher, militias and police violence

CONCLUSION

This first study in Brazil provides an understanding of the burden of FCS. Patients with FCS are affected by psychological, medical, and interpersonal problems, while the disease significantly impacts their socioeconomic life. **These findings support the need for:**

Understanding of burden of illness

More economic impact studies on the disease in Brazil.

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