Fragile X Syndrome Diagnosis and Patient Journey: The Caregiver’s Perspective

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BACKGROUND

• Fragile X syndrome (FXS) is a sex-linked genetic condition characterized by a range of developmental, neuropsychiatric, and psychiatric symptoms.
• Core FXS clinical symptoms include social avoidance/withdrawal, anxiety, irritability, deficits in learning and cognition, and sleep disturbances.
• FXS symptoms are frequently complicated by comorbid conditions, with attention-deficit/hyperactivity disorder occurring in an estimated 35% and 50% of male patients with FXS, respectively.

METHODS

• The survey was completed by 35 caregivers of children with FXS.

OBJECTIVE

• To characterize the patient journey in FXS surrounding diagnosis and clinical experiences through an online survey.

PARTICIPANTS

• Caregivers were predominantly female (80%), and most respondents were parents of children with FXS.

CURRENT EXPERIENCE OF FXS

• The most frequently experienced current symptoms of FXS were social avoidance and attention challenges, which were each reported by 85.7% of caregivers (Figure 5).

RESULTS

• Most caregivers (85.7%) rated the severity of FXS at the time of the survey as severe (45.7%) or moderate (40.0%) (Table 1a).
• The mean number of treatments children were currently receiving was 1.94.

PATH TO DIAGNOSIS

• The mean number of treatments children were currently receiving was 1.94.

CONCLUSIONS

• The mean number of treatments children were currently receiving was 1.94.
• The most common reason caregivers chose to enroll in the survey was to have a better understanding of FXS (54.3%) or to learn more about FXS (31.4%).
• Further research and education are needed to help caregivers gain early recognition of FXS by various stakeholders throughout the patient journey.

REFERENCES


Figure 1. Caregiver relationship to child with FXS.

Table 1a. Caregiver Demographics

<table>
<thead>
<tr>
<th>Demographic</th>
<th>Proportion of Respondents, %</th>
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<tbody>
<tr>
<td>Age</td>
<td>42.9 (28.7)</td>
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<tr>
<td>Male</td>
<td>57.1 (31.3)</td>
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<tr>
<td>Hispanic ethnicity</td>
<td>39.4 (24.6)</td>
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Figure 2. Most common factors prompting initial physician visit.

Figure 3. Length of time between noticing symptoms and initial physician visit.

Figure 4. First physician consulted.

Figure 5. Current FXS symptoms.

Figure 6. Current prescription medication received for FXS.