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Patients With Fragile X Syndrome Attending a Specialized Centre in Chile: Parent Satisfaction, Costs and Adherence

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Abstract

The study aim was to evaluate the administration and achievements of a Fragile X syndrome (FXS) centre in Chile. Families with children with FXS who sought care at our centre (CDTSXF) answered a survey to evaluate parent satisfaction. We analyzed adherence to management and the costs of CDTSXF operation. The percentage of parents satisfied with CDTSXF management ranged from 38% for speech development, to 100% for general management. The financial costs covered 70% of economic costs of centre operation. Patient adherence was associated with the age at admission to CDTSXF. Low satisfaction with language development could be attributed to low treatment adherence. The CDTSXF should study different mechanisms of financing its operation, estimate the cost of FXS for families, and improve treatment adherence among older patients.

Keywords: costs, Fragile X syndrome, practice implications, satisfaction, Treatment Adherence

Introduction

Fragile X syndrome (FXS, MIM #300624) is the most common form of inherited intellectual disability (ID) and autism spectrum disorders (ASD) in the world (Pugin et al., 2017), with an estimated prevalence ranging from 1.4 to 4 per 10 000 males and from 0.9 to 4 per 10 000 females (Hagerman, 2008; Hunter et al., 2014). Apart from these disorders, the cytosine-guanine-guanine (CGG) expansion in *FMR1* is also associated with a broad range of medical problems, both in patients with FXS and their relatives (Kidd et al., 2014; Pugin et al., 2017).

Chile is a South American country with almost 18 million inhabitants. The gross domestic product per capita is 15 732.31 U.S. dollars, but Chile has the worst income inequality among members of the Organization for Economic Cooperation and Development. Moreover, there are only 30 clinical geneticists in the country, far lower than the 170 recommended by the World Health Organization (Castillo Taucher, 2015). Thus, most of the population cannot be assessed for genetic disorders. Intellectual disability affects 1.9% of Chileans (National Fund for Disability, 2005), and although there is no estimated prevalence of FXS in Chile, near 8.2% ($n = 181$) of patients with ID studied in our laboratory over a 30-year period by *FMR1* polymerase chain reaction and/or Southern Blot have CGG triplet repeats >200 (Santa Maria et al., 2016).

Following the National Fragile X Foundation recommendations (National Fragile X Foundation, 2010), the Centre for Diagnosis, Treatment, and Monitoring of Patients with Fragile X Syndrome (CDTSXF) was created in 2011 at CEDINTA (Centro de Diagnóstico del Instituto de Nutrición y Tecnología de los Alimentos, an academic medical and research centre) to evaluate and follow patients with FXS and their families. It remains the sole centre dedicated to FXS in Chile. Briefly, CDTSXF is staffed by a clinical geneticist, a child neurologist (ChiN) and nutriologist, speech and occupational therapists (SpeT and OcT, respectively), and a family psychologist (FamPsy), who work together with a biochemist and cytogeneticists dedicated to molecular studies. The initial evaluation (IE) has six sessions; five of them are carried out for assessing the patient and her/his family's biomedical problems commonly seen in FXS. Finally, all the specialists' initial recommendations as well as parents' impressions are collected in the sixth session and delivered by the FamPsy and the chief of the centre to coordinate monitoring (Alliende et al., 2012). The CDTSXF also offers optional therapeutic sessions with medical specialists and a "treatment package" composed of eight speech and occupational interventions each and one session of follow-up with the ChiN (Alliende et al., 2012). The families

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can choose more than one "treatment package" and/or request follow-up appointments with medical specialists when necessary. Besides these activities, in the last 2 years our centre has developed an Annual OcT workshop and an Educational Seminar on FXS to improve the understanding of this disorder for education professionals.

The evaluation of a community healthcare system is a key step in improving its functioning in terms of maximizing the quality of the attention and users' satisfaction in a cost-efficient way (King's Health Partners, 2013; Saraceno & Levav, 1992). This is completely applicable to a mental care service, although it may be more difficult to assess an evaluation because some health parameters can be continuously evolving (Saraceno & Levav, 1992) and/or the community perception of well-being is not homogeneous among its individuals (Robert, Leblanc, & Boyer, 2015). For example, some parents of children with ID would be more satisfied with a service if they were involved in decisions about the intervention, whereas other parents might think that the best outcome for their children is addressed when professionals define the management (Robert et al., 2015). Conversely, the evaluation can vary depending on whom (e.g., family/caregiver vs. patient) is tested and how (e.g., structured vs. unstructured interview).

While it is recognized that people with ID should be assessed for satisfaction with healthcare support, there is currently no consensus on optimal measures of perceived satisfaction (Copeland, Luckasson, & Shauger, 2014); therefore, these individuals' families remain the main source of information. Saraceno and Levav (1992) have stated that an evaluation should consider four main points: three types of qualities (social, technical, and economic ones) and the efficacy of the intervention. The social quality refers to the acceptability and accessibility of the service for the community (e.g., the patients and caregivers/family satisfaction), the technical one denotes the implementation of updated and evidence-based management (e.g., following clinical guidelines), and the economic quality indicates the efficiency of treatment, that is, how many economic resources are invested to deliver a service. Finally, the efficacy of the intervention, or effectiveness, refers to the outcomes achieved in the practice (Saraceno & Levav, 1992). Following these recommendations, our aim was to evaluate the administration and achievements of a FXS centre in Chile.

Methods

Participants

All families with at least one child with FXS receiving treatment at the CDTSXF were invited to complete a survey, that is, 37 families and 39 patients. Patients' mean age at the time of the survey was 14.57 ± 7.55 years, and two patients were female (5.13%). Families were contacted by email, via the Chilean Fragile X Corporation, and in person when they brought their children for follow-up at the centre over a 4-month period in 2014. This study was approved by the Ethics Committee of Instituto de Nutrición y Tecnología de los Alimentos (INTA), University of Chile.

Measures

To our knowledge, there are no Latin American studies that evaluate the administration and achievements of a clinic devoted to FXS. Thus, the aims of our work were to describe parent satisfaction, costs and adherence of patients with FXS attending our specialized clinic as well as to examine the factors that influenced these outcomes, following the recommendations stated by Saraceno and Levav (1992) and taking into account some methodological aspects from the Ouyang, Grosse, Raspa, and Bailey (2010) study. The economic analysis considered a partial evaluation from the healthcare centre (the payer) perspective to calculate the cost of the interventions (Luyten, Naci, & Knapp, 2016). The Child and Adolescent Mental Health Clinical Academic Group outcomes book gives a good example on how to evaluate a service devoted to mental health and how evaluations improve management over time (King's Health Partners, 2013).

A web-based survey was developed to evaluate parent satisfaction about the treatment management that their children received at the CDTSXF (Appendix 1). Families completed one survey per child, thus, the maximum possible number of returned surveys was 39. Responders could be the patient's parents/guardians, a patient's close relative involved in her/his care, or the patient accompanied by a parent or caregiver. While the families of seven patients who lived outside of Santiago could complete the survey by email, the others could complete it by email or in person (see below). We emphasized that answering the survey was completely voluntary and anonymous, and their decision to complete the survey would not affect the quality/quantity of interventions given by the centre. We did not ask about responders' specific demographic data to maintain anonymity.

Briefly, the survey consisted of 42 multiple choice and 6 short answer questions, which were organized in 5 parts. The first part asked about age, gender, and types of evaluations carried out in our clinic; the second and third sections queried satisfaction with accessibility, facilities, and functioning of our centre in terms of punctuality, cost, schedule of sessions, and availability of specialists. The fourth part (goals of the program) asked about satisfaction with treatment provided by the health team, the medical indications, and evaluations—following the recommendations of the American Academy of Paediatrics (Hersh & Saul, 2011)—and the achievements reached by the patients. The last section inquired about general impressions of the CDTSXF.

The queries about level of satisfaction were questions with Likert type scale responses that ranged from "very unsatisfied" to "very satisfied," "very hard" to "very easy," or "very bad" to "very good." A "does not apply" option was available for every question. Lastly, the six short-answer questions were for patients' age and additional comments.

Appendix 2 shows the costs incurred by the health service associated with CDTSXF, including diagnosis and treatment pathways. The economic analysis was based on the comparison of economic and financial or budgetary costs over the period January 2011–December 2014. The former refers to all uses of resources, which have an economic ("opportunity") cost, whereas the latter refers to all financial expenses, which have a monetary cost. Costs were expressed in U.S. dollars (December 2014

exchange rate: US$1 = 612.92 Chilean pesos (CLP)). All costs were adjusted to 2014 prices and include the costs associated with clinical staff, laboratory examinations, overhead, and taxes (variable costs), plus operating costs and parenting advice activities (fixed costs). Operating costs include administrative expenses and salaries, whereas overhead is a function of outpatient income. Specialist consultations and laboratory examination costs were calculated according to the above-mentioned organization of CDTSXF. Staff costs associated with the diagnosis and examination were adjusted to account for overhead and taxes. Three sessions of parenting advice and education are provided via seminars and workshop to all patients and their parents at no cost. Medication is not provided by the program. The cost analysis of screening and treatment for depression in workplaces from the employer perspective shows how the cost of interventions was carried out for our centre (Evans-Lacko et al., 2016).

Finally, adherence was calculated for the Initial Examination (IE) and therapeutic appointments using computer registers of attendance of all patients treated at the CDTSXF. The total number of appointments carried out for IE was 205 and for therapeutic purposes was 865, from January 2011 to December 2014.

Procedures

The survey was delivered by email to each family when possible, which contained a link that redirected to the webpage with the survey. The survey was distributed by the informatics team at INTA, and created in and hosted temporarily by WordPress®. It was available from September 2014 to December 2014 and could be visualized and answered through personal computers, tablets, or smartphones. If our centre or the Chilean FXS Corporation (the organization of families with FXS) did not have an email for the family, then the survey was administered in person when the family brought the patient to the centre for follow-up. In this case, an independent person, not involved in analysing the data gave the survey to secure the anonymity. Both mechanisms of delivery contained the necessary instructions for independent completion. All families that attended our clinic were invited to participate.

Statistical Analysis

Survey results were expressed as numbers and frequencies in case of categorical or nominal variables, whereas continuous variables were described as means and standard deviations (SD). Adherence was calculated as a percentage (PoA) of the number of consultations carried out: total reservations/total number of expected sessions for each specialist, for the IE and management consultations. These were also expressed in medians and interquartile ranges (IR) or means and SDs when necessary. Adherence was also evaluated as a binary variable depending on if a minimum of 17 appointments was achieved or not.

We performed chi-square tests, simple logistic and ordinal regressions, Pearson's and Spearman's correlations to analyze the association between current age (CA), age under 12 years, age at admission to program (AAP), and every question of the survey, the PoA and number of appointments. The associations between type of evaluations and all questionnaire enquiries, as well as the access to our facilities were examined through the likelihood ratio or ANOVA tests. Finally, the relationship between residence and PoA and number of appointments were studied using the Mann–Whitney test, and the number of medical indications solicited vs. completed as well as PoA to IE vs. PoA to management were analyzed by paired t-test and Wilcoxon signed ranks test, respectively. A p-value < 0.05 was considered statistically significant.

Results

Survey

The descriptive survey results are depicted in detail in Appendix 3. The survey was completed by 18 families (18 patients in total) from a total of 37 families that are seen at the CDTSXF. Therefore, the overall response rate was 49%. The mean CA and mean age at diagnosis were 11.3 ± 4.6 and 6.0 ± 3.8 years, respectively, and all responder patients were male. The percentage of parents that declared to be "very satisfied" or "satisfied" with access to and CEDINTA's facilities, functioning of the FXS Program, the goals of the CDTSXF, and their general impressions ranged from 38 to 100%. The lowest level of satisfaction (near one third) related to the areas of speech, communication and language development, whereas 100% of families considered that CDTSXF was "beneficial" or "very beneficial."

There was no association between the CA and the goals of the program, excepting that pubertal stages were not explored in patients under 12 years whereas this variable was examined in 57.1% of patients $\geq$12 years ($p = 0.026$). On the other hand, there were no associations between the kind of evaluation received and the goals of CDTSXF.

Economic Analysis

While the economic costs considered all the resources delivered by the CDTSXF, the financial costs analysis took into account all the monetary expenditures CDTSXF carried out. Under the current strategy, the differential between economic and financial costs equalled 28.7% of overall economic costs (Table 1). The major contribution to costs came from expenses in support of the organization rather than to the clinical program (45.2% of economic costs or 63.4% of financial costs). Salaries for clinical staff accounted for less than 10% of financial costs because most of the clinicians perform voluntary work, however, when economic costs were taken into account their contribution to the overall costs increased to 22.2%. Financial expenses of diagnosis and treatment accounted for approximately one third (36 and 30%, respectively) of economic costs.

Adherence to CDTSXF

Concerning the adherence of patients with FXS attending CDTSXF ($n = 39$), all specialties had over 74% attendance for the IE, but this percentage decreased for on-going management to a global percentage of 33.3%, with the highest attending rate for the ChiN (74.2%) and the lowest for sessions with SpT

TABLE 1
Economic vs. financial costs of the CDTSXF, in US$

	Economic costs			Financial costs		
Concept	**Variable costs**	**Fixed costs**	**Total costs**	**Variable costs**	**Fixed costs**	**Total costs**
Salary (clinical staff only)	20 371.1		20 371.1	6 330.5	. . .	6 330.5
Laboratory examinations	14 887	. . .	14 887	14 268.7		14 268.7
Medicines and drugs	NP	. . .	NP	. . .	NP	NP
Operating costs[a]	. . .	41 530.7	41 530.7	. . .	41 530.7	41 530.7
Overhead costs	7 835	. . .	7 835	2 434.8	. . .	2 434.8
Taxes	3 134	. . .	3 134	973.9	. . .	973.9
Parenting advice and education	. . .	4 144.1	4 144.1	. . .	0	0
Total	**46 227.1**	**45 674.8**	**91 901.9**	**24 007.9**	**41 530.7**	**65 538.6**

Exchange rate: US$1 = CLP 612.92 (December 2014).

[a]Operating costs include administrative expenses and salaries.

NP, not provided by CDTSXF.

(36.7%). Figure 1 depicts the main associations found between adherence and number of consultations with regard to different variables. As can be seen, the median adherence to IE sessions (median 100%, IQR 83–100%) was significantly higher than follow-up sessions (median 73.5%, IR 29.3–90.1%, $p < 0.001$). Conversely, AAP predicted the adherence to the delivery session of IE, the adherence to management by ChiN, and the number of treatment consultations. No other variables (e.g., city of origin) were found to be associated with adherence and number of appointments.

Discussion

We presented the evaluation of a specialized rare disease clinic, and were unable to compare our findings with a similar study because to our knowledge this type of analysis has not been described previously. Although there are several articles that show the economic burden and family impact of FXS (Angelis, Tordrup, & Kanavos, 2015; Bailey et al., 2012; Reilly, Murtagh, & Senior, 2015; Sacco, Capkun-Niggli, Zhang, & Jose, 2013) and at least one article that describes the impact of a specialized health programme for children with FXS in the United States (Hatton et al., 2000), there are no similar articles from Latin America. We analyzed parent satisfaction with the management of their children with FXS and their adherence to treatment indications. Moreover, no other study has shown results from the medical centre's perspective, which is essential to diminish centre costs, improve parent satisfaction, and patient adherence.

There was considerable parent satisfaction with the CDTSXF, but satisfaction decreased when the therapeutic achievements were analyzed, specifically in areas of the speech, communication, and language. This may be attributed to FXS itself and as part of the challenges that parents of children with FXS face (Reilly et al., 2015). It is well-known that 22% of patients with FXS have ASD (Richards, Jones, Groves, Moss, & Oliver, 2015) and 80% of individuals with this genetic disease have attention deficit-hyperactivity disorder (Hersh & Saul, 2011) besides ID as a comorbid condition. Moreover, families with children who have ASD and other disorders tend to be less satisfied with the healthcare system, reporting lower doctor satisfaction, and longer delays in care (Zablotsky, Kalb, Freedman, Vasa, & Stuart, 2014). While our survey did not ask about other clinical diagnoses such as ID, ASD, and others, we hypothesize that the lower levels of satisfaction in therapeutic achievements are related to the comorbid disorders in these patients. Conversely, our results about parent satisfaction coincided with those reported by Hatton et al. (2000), who describe a high general satisfaction with the intervention for their children, although parents reported a preference to increase the amounts of speech–language and occupational therapies. Thus, satisfaction among Chilean parents seems to be similar to other populations of parents of children with FXS.

As was discussed above, the lower satisfaction with speech and communication development may be explained by FXS *per se*. However, we cannot discard the effect of low adherence to appointments with the SpeT in parent satisfaction levels with therapeutic achievements in this area. Indeed, an Australian study showed that parent satisfaction with the community service for people with ID was directly proportional to the frequency of occupational therapy sessions (Wilkins et al., 2010). Moreover, adherence and the request for therapeutic consultations were strongly determined by the patients' AAP, which is concordant with the lower use of therapeutic services seen in these patients at higher ages (Martin et al., 2013). Considering this evidence, we could hypothesize that older patients require less health attentions because they have achieved some skills (e.g., ability to communicate and socialize sufficiently) and therefore, the family did not deem it necessary to assist with speech or occupational therapy appointments. Other explanations for the lower adherence seen in older participants with FXS are: (1) families dissatisfied with the management given and its achievements; (2) patients required less health attention because parents have become discouraged over time; (3) families have pressing domestic circumstances; (4) they are more poor; or (5) parents suffer greater

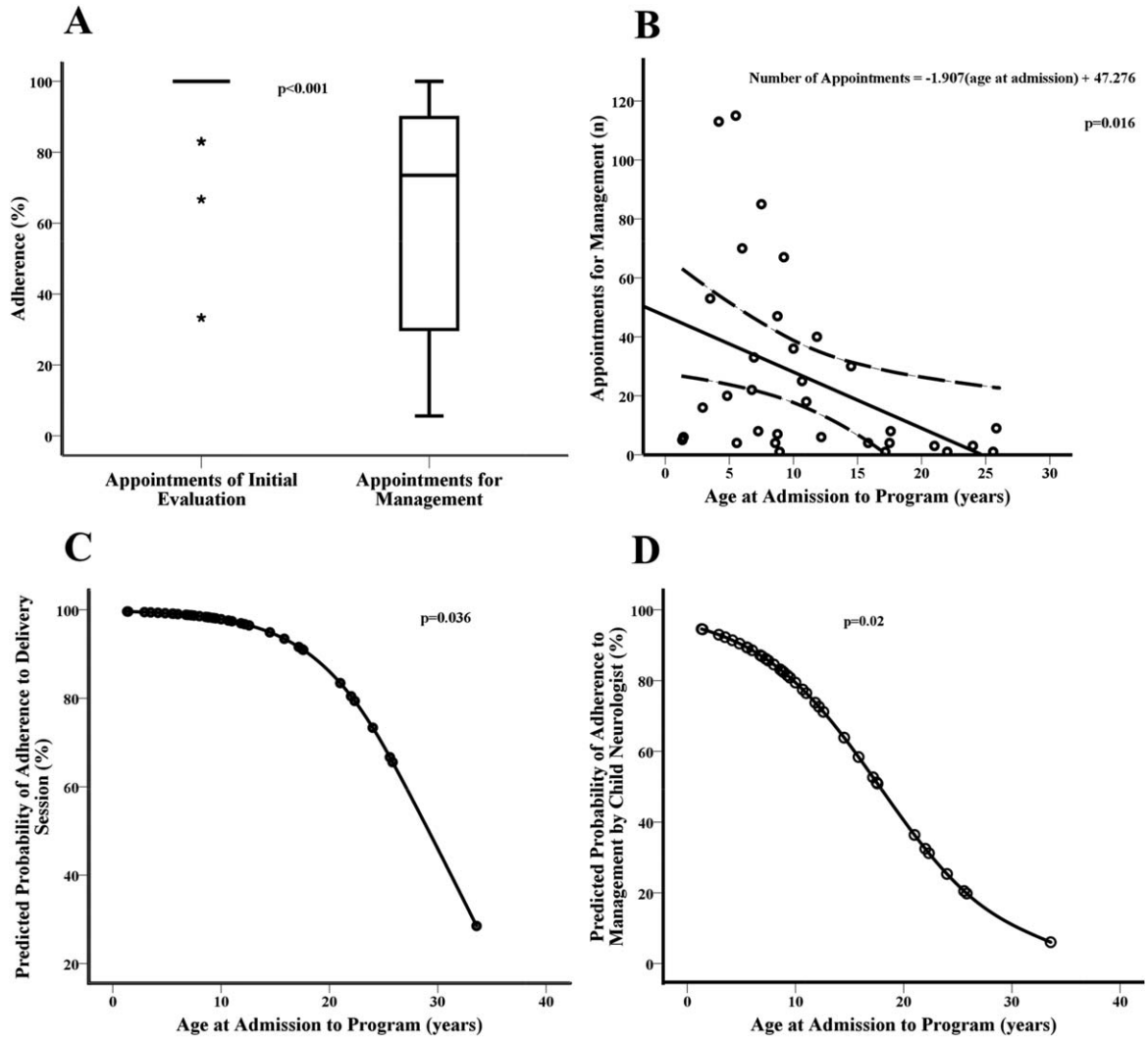

FIGURE 1

Associations found between adherence and number of consultations with regards to the type of evaluation and age at admission to program (AAP). (a) Adherence for Initial Evaluation (IE) sessions was significantly higher than that of follow-up appointments. The AAP predicted the total number of consultations for management (b), the attendance to the delivery session of IE (c), and adherence to management by a child neurologist (d).

financial burdens. Nevertheless, the literature about this issue is scarce to support these conclusions and our questionnaire was not designed to provide answers to these questions, but rather to describe an unknown situation. Despite this, our centre should carry out a plan to improve the treatment adherence of older patients.

Regarding our economic analysis, the CDTSXF partially subsidized the costs of molecular diagnosis and therapeutic sessions due to nonexistent coverage of health insurance for rare diseases in Chile. Whether it is also considered that overhead rates imposed by CEDINTA had to be discounted from incomes, these two factors may explain the high CDTSXF operating costs and, therefore, the great difference between economic and financial costs. Future work should determine if our subsidy policies are adequate and should explore other ways of financing to not increase the families' out-of-pocket expenses. This last point is a key factor to keep in mind by the professionals who work with patients with FXS because it has been well demonstrated that FXS is one of the most expensive genetic diseases (Angelis et al., 2015). Parents of children with FXS have to reduce their employment significantly (Ouyang et al., 2010) and have a greater financial burden compared with parents with children with ASD only

(Ouyang et al., 2014). These issues should be studied in our population to estimate the overall cost of FXS and the effectiveness of our interventions, that is, to perform a full economic evaluation (Luyten et al., 2016). This could help us to find better ways of ameliorating the patients' quality of life.

The main limitations of this study were its small sample (18 families, 39 patients), which could explain why there were not more significant associations, and its low response rate. However, it is difficult to expand the sample for several reasons. First, Chileans living with ID are not usually assessed for FXS due to genetic examination costs and lack of clinical geneticists and well-trained clinicians that might suspect FXS. Second, individuals from Chilean regions outside of the capital, Santiago, cannot access our centre easily. Finally, Chile has a small population and no current laws that can help to support CDTSXF.

The CDTSXF along with the Chilean FXS Corporation have been recently developing conferences for both the healthcare community and school professionals outside the capital city to teach about the syndrome to improve the diagnosis of FXS in patients with ID. This may also generate collaborations with researchers in other parts of the country to perform large-scale studies and try to replicate this type of centre in isolated communities. Furthermore, the Chilean FXS Corporation is evaluating mechanisms to financially assist families with difficulties in testing other at-risk relatives. We think that these activities and the present work may guide similar centres in developing countries to evaluate their processes and functioning to improve services provided to patients.

Conflict of Interest

The authors declared no potential conflicts of interest with respect to the research, authorship and/or publication of this article.

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Supporting information

Additional Supporting Information may be found in the online version of this article at the publisher's website.

APPENDIX 1 Survey.

APPENDIX 2

FIGURE S1 Diagram showing variable and fixed costs of the CDTSXF.

APPENDIX 3 Whole results of survey. All participant patients were males.

TABLE S1 Identification and type of attention ($N = 18$; mean age $\pm$ SD= 11.3 ± 4.6 years).

TABLE S2 Access and CEDINTA's facilities ($N = 18$; mean age $\pm$ SD= 11.3 ± 4.6 years).

TABLE S3 Functioning of Fragile X syndrome program ($N = 18$; mean age $\pm$ SD= 11.3 ± 4.6 years).

TABLE S4 Level of Satisfaction with the Professional-Family relationship.

TABLE S5 Evaluations and Indications.

TABLE S6 Therapeutic achievements ($N = 18$; mean age $\pm$ SD= 11.3 ± 4.6 years).

TABLE S7 Nutritional evaluation.

TABLE S8 General impressions ($N = 18$; mean age $\pm$ SD= 11.3 ± 4.6 years).