**Healthcare system and professionals and SXF- The families perspectives**

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**ABSTRACT**

Introduction: The Fragile X Syndrome is a developmental disorder with genetic etiology not very common and often underdiagnosed in Portugal.

However, it is the most frequent inherited cause of intellectual disability and also the better known genetic origin of autism. Health professionals are those who, firstly, are responsible for detecting the signs that something is affecting the development and conduct to the diagnosis.

Methods: Qualitative study based on interviews with 40 mothers of children with FXS (full mutation) using a Grounded Theory approach.

Results: We found: a) difficulty of pediatricians in dealing with complaints and signals detected by the mothers, b) little information or awareness of referral for diagnosis; c) significant delay in sending for genetic diagnosis d) difficulties in communicating the diagnosis to families, e) articulation and difficulties monitoring throughout the development of the child.

Conclusions: Based on these findings will be proposed guidelines so that the health system can receive and follow more effectively the children and families living with this syndrome.

**Keywords** – Fragile X syndrome, diagnosis, disability

***Supplementary Information to Provide:***

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