

ABSTRACT

The thesis deals with the family of a child with Kabuki syndrome. In the Czech Republic, the diagnosis was established in 2010. The main objective of the thesis was to compile comprehensive information about the family of a child with Kabuki syndrome and to determine more detailed information about the functioning of this family in the areas of upbringing, experiences with the school system, and also the utilization of support by the family. The thesis is divided into theoretical and empirical parts. The theoretical part is divided into four chapters. The first chapter discusses the definition of Kabuki syndrome, the history of research on this rare condition, diagnosis, and characteristic features in individuals with Kabuki syndrome. The second chapter focuses in more detail on the clinical symptoms of Kabuki syndrome, from health issues and treatment options to levels of cognitive function. The third chapter specifies on families of children with rare diseases, and the following chapter discusses the possibilities of support for families of children with Kabuki syndrome. The empirical part consists of a research investigation focusing on a child with Kabuki syndrome, the family of the child, and the functioning of the family through semi-structured interviews and a case study.