

Abstract

The thesis is about a family of a child with Kabuki syndrome. The first chapter deals with the syndrome itself, diagnosis, clinical symptoms and cognitive functions. The second chapter describes the family with the rare condition, the family in the context of the history and what stages the family of the child with the rare condition goes through. The next chapter discusses support for the family with a rare disease, and also includes the concept of a non-profit organization and an association. The practical section explores the family of an individual with Kabuki syndrome, and includes an interview with a family member. The research section is supplemented with a case study.