Long-term support for people with genetic conditions
- The role of the certified genetic counselor -

KANBARA Yoko

Social welfare and other support systems to aid individuals with childhood genetic diseases are generous in Japan. On the other hand, those who apply during adulthood receive less support, and self-application to the local government is required to obtain continuous assistance. Parents and caregivers of children with genetic diseases must obtain information about the available support system and start the application process before the children reach adulthood. If parents and caregivers do not have the opportunity to access information, they cannot apply for welfare in adulthood. Support for genetic diseases requires information about the disease as well as about welfare. Down syndrome (DS) is one of the most common genetic conditions. Although information about DS in childhood is abundant, knowledge about adult DS is limited and the condition is not well understood, even by healthcare professionals. In the case of less frequent and rare diseases, such as the cardio-facio-cutaneous (CFC) and Costello syndromes, even when a diagnosis is made, the absolute amount of information about the disease is scarce. In recent years, diagnostic programs and databases linked to government programs for intractable diseases have been established for undiagnosed patients based on clinical information and gene analysis data. The interpretation of genome sequencing data is complex, and data handling requires consideration of the characteristics of the genetic information. With the increase in information associated with genetic diseases, patients and their parents may face difficulties understanding the relevant data. Genetic counseling is necessary to achieve this understanding. However, details regarding support provision remain elusive. This thesis aimed to clarify the actual situation of information provision and propose a role for a certified genetic counselor (CGC) by examining the effectiveness of the involvement of a CGC, a genetics professional, as a supporter of individuals with genetic conditions and their families.

This thesis comprised three studies. In the first study, a retrospective survey of medical records was conducted on adults with DS who visited the outpatient clinic of the internal medicine department of a rehabilitation medical center in Japan. The aim of the study was to clarify the health problems of adults with DS. The results showed that endocrine disorders were common at all ages, neurological disorders were common in patients over 41 years, and patients in their 20s and younger had many comorbidities that increased with age. Therefore, continuous medical care is necessary for the prevention and early detection of comorbidities in patients with DS. Additionally, most middle-aged patients with DS lived with their families, and approximately half were employed in welfare facilities. The reasons for visiting a clinic varied, with the largest number of visits being for symptomatic conditions, while approximately 30% of the visits were for medical checkup examinations without overt symptoms. Larger scale surveys are needed to ascertain the actual situation in middle-aged and older adults with DS.

In the second study, a seminar on DS in adulthood was organized to provide information to adults with DS and their families. The participants responded to a questionnaire survey regarding the provision of information and family support. The results revealed that parents of children with DS were highly concerned about cognitive function, dementia, and emotional and behavioral disorders in adulthood. Importantly, satisfaction with the seminar was very high, and all participants expressed interest in attending subsequent seminars. The seminars conducted by the medical institutions served as a source of information on DS desired by the families and can be useful sources of information in the future.

The third study proposed the support of genetics professionals for patients with intractable and rare diseases through group clinics and roundtable discussions on the CFC and Costello syndromes, which affect a small number of patients. However, as the history and comorbidities of these syndromes have not been well described, the author attempted to understand medical and life-related problems from the narratives of parents' experiences in raising their children. The transcripts of the roundtable discussions were analyzed using the Steps for Cording and Theorization (SCAT) method. According to the SCAT analysis, the parents' narratives included difficulties in parenting due to condition-specific symptoms, encouragement and future prospects for others, and empirical narratives with a powerful message. The content suggests that genetics professionals should play an educational role in addressing the difficulties faced by people with rare diseases and in providing disease information and care to healthcare professionals.

In summary, the role of CGCs in providing information to people with genetic conditions and their parents includes (1) providing information on social welfare and medical subsidies, (2) providing disease information for the health care of people with genetic conditions, (3) providing support to those who have not received genetic counseling, and (4) providing information to the general community, including patients and their families. People with genetic conditions must be provided with information on social welfare and health care subsidies. Furthermore, since lifelong consultations are important for people with genetic conditions, motivating patients/families to continue receiving medical care and to manage their own health is important. CGCs must act as hubs to achieve team medicine involving multiple professions and establish a broad network with other departments and institutions. In addition, CGCs should play a role in educating other healthcare professionals. Genetic conditions require lifelong health care, and CGCs need to demonstrate that they are up-to-date and well-informed as medical professionals and supporters who are continuously involved in the growth and development of the individual from childhood to adulthood. In recent years, comprehensive genomic analyses have revealed the causes of disease. Due to the nature of this genetic information, understanding its content is difficult for patients and their families, and therefore genetic counseling is necessary. Thus, long-term patient support requires the input of several genetics professionals. Further education to understand disease requires national and society-wide efforts.