Brief Communication

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Quality of Life in Prader-Willi Syndrome Patients

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Prader-Willi syndrome (PWS), a complex multisystem genetic disorder, is caused by the lack of expression of paternally inherited imprinted genes on chromosome 15q11-q13[1]. PWS has various characteristic features, including neonatal hypotonia, developmental delay, short stature, behavioral abnormalities, childhood-onset obesity, hypothalamic hypogonadism, and characteristic appearance[2]. The course and early natural history of PWS patients can be divided into two generally recognized clinical stages. Symptoms of PWS include infantile hypotonia and failure to thrive in the neonatal and early infancy period, followed by life-long hyperphagia, developmental delays, moderate-to-severe behavioral problems, and several physical problems that impact health.

Regarding on the hyperphagia period, PWS patients are unable to control food intake and will overeat if not monitored. Their food-seeking behaviors and low metabolic rate make them obese. Obesity is the greatest threat to their health, and these patients often die young from obesity-related complications. While no medications are currently known to aid in controlling hyperphagia, human growth hormone is used for the treatment of PWS because it can help normalize height, increase lean body mass and mobility, and decrease fat mass[3].

Other symptoms associated with PWS usually include cognitive challenges and learning disabilities, growth hormone deficiency and short stature, small hands and feet, scoliosis, sleep disturbances with excessive daytime sleepiness, high pain threshold, difficulties with speech and movement, and infertility. Behavioral difficulties may include obsessive-compulsive symptoms, skin picking, and difficulty controlling emotions. Treatments for many of these secondary complications include education planning, speech therapy, and growth and sex hormone replacement therapy. Serotonin reuptake inhibitors (SRIs) are also helpful for most teenagers and adults in managing mood-related symptoms.

The complex genetics, etiology, multiple phenotypes, and evolving natural history of PWS suggest that a multidisciplinary professional, parental, societal, and environmental approach to the management of these patients is required for overcoming the many challenges of reducing morbidity and mortality and improving patient quality of life (QoL)[1]. The multidisciplinary team involved in the care of PWS patients and their families should include several medical and non-medical professional figures (e.g. child psychiatrists, pediatric endocrinologists, clinical geneticists, orthopedic specialists, educational psychologists, and social workers). However, all these professional figures should know what the patient wants and what kind of relationship there is between the traditional clinical outcome measures and the patient's perspective[4]. Over the last two decades, clinical researchers have emphasized the need for a thorough evaluation of the patient's QoL in order to study the impact of chronic illnesses and their treatments on the patient's life[5]. The need for standardized measures of such items stimulated an extensive and rigorous process allowing the development of validated patient-oriented instruments[6].

There are few reports on QoL in patients with PWS. Caliandro et al. reported an assessment of QoL in patients with PWS in Italy[4]. They performed a multi-center study on 40 consecutive patients with PWS evaluated through the Short Form-36 and the Child Health Questionnaire-Parent Form-50 according to the age of patients, which showed intensely impaired QoL in mental and physical aspects. Weight at the moment of observation and

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characteristic facial features are the main variables that influence QoL.

Furthermore, families/mothers/siblings of PWS patients showed poorer perceived QoL[7]. The level of parental stress and pessimism associated with PWS was higher than that in families of children with intellectual disabilities of mixed etiologies. Mothers and unaffected siblings also need psychosocial support due to the multiple challenges of living with and caring for a child/young adult with PWS.

In conclusion, PWS is a genetic disorder characterized by extreme obesity accompanied by other multi-system clinical manifestations encompassing both physical and behavioral/cognitive abnormalities. Therefore, the systemic approach of a multidisciplinary team should be involved in the care of PWS patients and their families, and thorough assessment of multiple aspects should be essential for improvement of their QoL. To ensure better QoL for patients with PWS in Korea, there is a need to make a proper standardized measurement for Korean PWS patients.

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