Down Syndrome

Subjects: Pathology

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Down Syndrome (DS) (OMIM#190685, Online Mendelian Inheritance in Man®, An Online Catalog of Human Genes and Genetic Disorders) is a genetic disorder caused by a trisomy of chromosome 21 and is the most common genetic cause of intellectual disability (ID).

Keywords: Down's syndrome ; Trisomy 21 ; intellectual disability ; overweight ; obesity ; aging

1. Introduction

DS is associated with significant health problems as diseases such as congenital heart disease, obstructive sleep apnea, celiac disease and endocrinopathologies. Endocrine disorders are usually characterized by thyroid disorders, low bone mass, diabetes, short stature and propensity to be overweight/obese[1][2].

2. Impact

Life expectancy of people with DS has increased significantly from 12 years in 1949, to 60 years in 2004, and it is expected to increase in the near future[1][2]. Unfortunately, the increase in life expectancy is not parallel to the increase in the period of life with optimal health. Accelerated aging is identified in the case of DS based on two aspects: a. Clinical-pathological characteristics of the subject; b. Monitoring of molecular markers related to biological age and the aging process, highlighting the shortening of the telomeres[1] . Thus, several studies have connected the shortening of the telomeres with obesity, and particularly with the increase of BMI and adiposity causing accelerated aging[3][4].

Currently, there is a higher prevalence of overweight in ID patients (≥18 years) compared to those not affected by IDs, both in obesity (38.3% vs, 28%) and in morbid obesity (7.4% vs, 4.2%)[5]. Prevalence in overweight and obesity varied between 23–70% in DS patients (13.3–52.9 and 0–62.5%). Thus, young people with DS have higher rates of overweight and obesity than young people without DS[6].

The causes of the development of overweight and obesity in DS are: hypotonia (decreased muscle tone), susceptibility to systemic inflammation, metabolic diseases and / or slow metabolism[7]. Usually, people affected by DS consume less healthy food, and show physical limitations, depression, and lack of social and financial support. Besides, medications contribute to weight gain[8]. The key challenge for this field of knowledge in incoming years will be to identify therapeutic intervention strategies for weight loss that reduce body fat and systemic inflammation[6][9]. Therefore, there is a need to increase evidence-based clinical knowledge, with the aim of improving existing care programs[2][10][11].

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