Rev Med Int Sindr Down. 2016;20(2):17–20

FUNDACIÓ CATALANA SÍNDROME DE DOWN
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INTERNATIONAL MEDICAL REVIEW ON DOWN'S SYNDROME

www.elsevier.es/sd

ORIGINAL

Pulmonary function in young adults with Down syndrome: A cross-sectional study

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Received 14 April 2015; accepted 14 May 2015
Available online 17 July 2015

KEYWORDS
Down syndrome;
Lung function;
Body mass index

Abstract
Aim: To compare the pulmonary function of young adults with Down syndrome (DS) with healthy subjects.
Methods: Thirty-four young adults (17 with DS and 17 apparently healthy controls), aged 20–40, participated in this study. Anthropometric variables and lung function, namely forced expiratory volume in one second (FEV_1), forced vital capacity (FVC), peak expiratory flow (PEF), and the fraction of FVC expired in one second (FEV_1/FVC%), were assessed in both groups.
Results: The group of young adults with DS had a lower height and higher body mass index (31.4 ± 4.6 vs. $23.4 \pm 1.3\,kg/m^2$, $p < 0.001$). As regards pulmonary function, the group of participants with DS showed significantly lower values for PEF (238.4 ± 89.4 vs. $387.4 \pm 52.9\,L/min$, $p \leq 0.001$), FVC (2.2 ± 0.7 vs. $3.1 \pm 0.4\,L$, $p \leq 0.001$) and FEV_1 (1.9 ± 0.6 vs. $3.1 \pm 0.5\,L$, $p \leq 0.001$), when compared to subjects of the control group. No changes were observed in FEV_1/FVC%. An inverse correlation was observed between the body mass index and the PEF ($r = -0.691$, $p < 0.001$), the FVC ($r = -0.555$, $p = 0.001$), and the FEV_1 ($r = -0.617$, $p < 0.001$).
Conclusion: Young adults with DS showed reduced pulmonary function in comparison to age-matched controls. Additionally, the pulmonary function was inversely correlated with body mass index.

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http://dx.doi.org/10.1016/j.sdeng.2015.05.004

PALABRAS CLAVE
Síndrome de Down;
Función pulmonar;
Índice de masa corporal

Función Pulmonar en adultos jóvenes con Síndrome de Down: Estudio Transversal

Resumen
Objetivo: Comparar la función pulmonar entre adultos con Síndrome de Down (SD) y adultos sanos.
Método: Participan en el estudio 34 adultos jóvenes (17 con SD y 17 controles sanos) con edades comprendidas entre los 20 y 40 años. Se registran en ambos grupos variables antropométricas y de función pulmonar: volumen espiratorio forzado en un segundo (FEV_1), capacidad vital forzada (FVC), flujo espiratorio máximo (PEF) y la relación FEV_1/FVC.
Resultados: El grupo con SD presentaron un mayor peso e índice de masa corporal (IMC) que el grupo control ($31,4 \pm 4,6$ vs. $23,4 \pm 1,3$ kg/m2, $p < 0,001$). Se registraron valores significativamente menores en la función pulmonar de los sujetos con SD que en la del grupo control: PEF ($238,4 \pm 89,4$ vs. $387,4 \pm 52,9$ L/min, $p \leq 0,001$), FVC ($2,2 \pm 0,7$ vs. $3,1 \pm 0,4$ L, $p \leq 0,001$) and FEV_1 ($1,9 \pm 0,6$ vs. $3,1 \pm 0,5$ L, $p \leq 0,001$). No se observo diferencias entre grupos en el FEV_1/FVC. Se observó una correlación inversa entre el IMC y el PEF ($r = -0,691$, $p < 0,001$), la FVC ($r = -0,555$, $p = 0,001$) y la FEV1 ($r = -0,617$, $p < 0,001$).
Conclusión: Los adultos con SD muestran una reducción de la función cuando se les compara con controles de su misma edad. La función pulmonar correlaciona inversamente con el IMC.

Introduction

Down syndrome (DS) is characterised by multiple malformations, cognitive impairment and medical issues due to the presence of extra genetic material from chromosome 21.[1,2] Regardless phenotype variability, there are multiple common features that contribute to establish a diagnosis of DS such as hypotonia, small brachycephalic head, epicanthal folds, flat nasal bridge, Brushfield spots, small mouth, small ears, and excessive skin at the nape of the neck, among others.[1,2] Children and adolescents with DS present a combination of mental retardation, that is variable from mild to severe, with neuromuscular impairment, such as muscle hypotonicity, hypermobility of the joints or ligament laxity, light to moderate obesity, characteristic facial features, congenital heart disease, immunological dysfunction, hypothyroidism, pulmonary hypoplasia, visual and auditory problems, poor balance, perceptual difficulties, and other health problems.[1,3]

The pulmonary hypoplasia in children with DS results in lung growth abnormalities, such as fewer terminal lung units, acini with reduced number of alveoli, spacious and distended alveolar ducts, and smaller alveolar surface area.[4] Also, lower respiratory tract infections occur more commonly in children with DS, and these combined with respiratory muscle weakness, may result in functional respiratory impairment.[3,4]

The assessment of pulmonary function is of great importance, as it could be used to assess the baseline respiratory function, to detect pulmonary problems, and to monitor the effectiveness of rehabilitation strategies. Additionally, it could help the definition of intervention strategies in order to improve respiratory muscles strength, thus improving lung compliance and assisting the prevention of other negative phenomena associated such as secretions retention, decreased lung volumes, recurrent lung infections and decreased effectiveness of cough. Despite being a simple, inexpensive, feasible and quantifiable measurement, few studies investigated the pulmonary function in persons with DS.[5–8] Thus, the aim of the present study was to compare the pulmonary function of young adults with DS with healthy subjects.

Materials and methods

Participants

A total of 34 young adults (20 females and 14 males) with an age range between 20 and 40 years voluntarily participated in this cross-sectional study. This convenience sample was composed by 17 apparently healthy young adults (9 females and 8 males) and 17 young adults with DS (11 females and 6 males). The group of individuals with DS was recruited at two special education and professional training centres localised in the Oporto area, Portugal, after authorisation to perform the study has been granted by the participants, their parents and/or guardians and the centres' director. Each participant's parents/guardians were asked to complete a questionnaire detailing their relative medical history. The inclusion criteria were: adults with moderate to mild mental retardation, able to walk independently, without serious visual and/or auditory problems, and with medical clearance from the participant's physician to participate in the study. Exclusion criteria were as follows: associated congenital heart abnormalities, participating in sport activities, neuromuscular or orthopaedic disorders involving the thorax, including upper respiratory tract infection or back pain within the three weeks prior to data collection. In addition, a physiotherapist examined the participants to determine whether any of them had any spine or chest wall deformities