Clinical and Cardiorespiratory Assessment in Children With Down Syndrome Without Congenital Heart Disease

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Objective: To assess the clinical and functional status of a young Down syndrome (DS) population without congenital heart disease.

Design: Prospective study of children with DS and control subjects.

Setting: Bambino Gesù Children’s Hospital, Rome, Italy.

Participants and Methods: Forty-two children with DS (mean ± SD age, 9.8 ± 3.6 years) underwent genetic, clinical, neuropsychological (IQ), and cardiorespiratory evaluation. Cardiorespiratory fitness was assessed with a treadmill test and a lung function test to determine forced vital capacity, first-second forced expiratory volume, and peak expiratory flow. Data were expressed as a percentage of the predicted values for control children. To assess cooperation during exercise testing, we devised a compliance scale assigning a score according to the subjects’ ability to understand instructions, ability to walk and run as required, and need for vocal encouragement.

Results: Eighteen (43%) of 42 children with DS were obese, 10 (24%) were short, and 17 (40%) had microcephaly. On the Leiter International Performance Scale, 2 of 35 subjects had a normal IQ score (80-120); all others had low IQ scores (79 to <40). Five subjects did not undergo cardiorespiratory assessment. Eighteen of 37 subjects completed lung function tests: the results for 10 were unremarkable, and results from 8 revealed reduced forced vital capacity because of poor compliance. The subjects tested had low exercise tolerance (mean ± SD tolerance percentage, 61% ± 12%), mild tachycardia (maximal heart rate, 91% ± 4%), and a mild hypertensive response (maximal blood pressure, 89% ± 8%). Compliance scores correlated significantly with exercise time and age but not with IQ.

Conclusions: Clinical and cardiorespiratory assessment is feasible in subjects with DS without congenital heart disease and should be useful in gauging their fitness level for safe physical activity.


Editor’s Note: This study should be of special help to those involved in the Special Olympics—which should be all of us.

Catherine D. DeAngelis, MD

The incidence of Down syndrome (DS) is 1 in 700 to 800 births. About 40% to 50% of people with DS have congenital heart disease. In general, the most common anomalies are atrioventricular canal defect, tetralogy of Fallot, and patent ductus arteriosus. The severity of the underlying congenital heart disease adversely affects survival. People with DS without congenital heart disease survive relatively longer (about 90% longer) than those with congenital heart disease. The outcome of those without congenital heart disease depends primarily on the secondary manifestations of DS, namely mental retardation and musculoskeletal disability. The purpose of this study was to assess the clinical and functional status of a population of children with DS without congenital heart disease who sought permission to engage in regular physical activity.

Results: Eighteen (43%) of 42 children with DS were obese, 10 (24%) were short, and 17 (40%) had microcephaly. On the Leiter International Performance Scale, 2 of 35 subjects had a normal IQ score (80-120); all others had low IQ scores (79 to <40). Five subjects did not undergo cardiorespiratory assessment. Eighteen of 37 subjects completed lung function tests: the results for 10 were unremarkable, and results from 8 revealed reduced forced vital capacity because of poor compliance. The subjects tested had low exercise tolerance (mean ± SD tolerance percentage, 61% ± 12%), mild tachycardia (maximal heart rate, 91% ± 4%), and a mild hypertensive response (maximal blood pressure, 89% ± 8%). Compliance scores correlated significantly with exercise time and age but not with IQ.

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SUBJECTS AND METHODS

TESTING MEASURES

We studied 42 subjects, 25 boys and 17 girls, referred to our hospital for periodic clinical follow-up (mean ± SD age, 9.8 ± 3.6 years; age range, 4-18 years; weight, 33 ± 13.7 kg; and height, 128.3 ± 17.3 cm). All of the subjects had DS without congenital heart disease and underwent genetic (chromosome analysis), clinical (anthropometry), neuropsychological (IQ according to the Leiter International Performance Scale4), and cardiorespiratory assessment. They also underwent an echocardiographic examination to exclude cardiac disease. Subjects underwent a maximal Bruce Treadmill Test† with a 12-lead electrocardiogram to investigate exercise time (duration of exercise in minutes), maximal heart rate (beats per minute), and maximal systolic blood pressure (millimeters of mercury, palpatory measurement with an aneroid Tycos sphygmomanometer [Taylor Instrument, Asheville, NC]). Fifteen minutes after stopping exercise, subjects underwent a lung function test to determine forced vital capacity, first-second forced expiratory volume, and peak expiratory flow rate (liters per minute). All exercise data are expressed as a percentage of predicted values obtained in our laboratory for a healthy, age- and sex-matched population of nonathletic children.6 We also compared maximal heart rate and maximal systolic blood pressure (at peak exercise time) in subjects with values obtained for controls at the same exercise time.

Because exercise testing yields reliable results only when subjects collaborate, to assess compliance for subjects with DS, we devised a scale comprising 3 items: (1) ability to understand the instructions given during testing; (2) ability to perform the necessary actions, including coordinated walking and running; and (3) the need for vocal encouragement. Two physicians (E.P. and A.T.) assigned subjects a total score (1) ability to understand the instructions given (score, 1); 2, poor understanding (score, 2); 3, reasonable understanding (score, 3); 4, good understanding; and 5, excellent understanding. Early dementia symptoms were an exclusion criteria for exercise testing.

Children’s parents completed a questionnaire eliciting information regarding their child’s regular physical activities and gave written informed consent for their children to participate in the study.

STATISTICAL ANALYSIS

Values are expressed as mean ± 1 SD. Linear regression analysis was used to correlate compliance scores with exercise time, age, and IQ scores. The t test was used to compare maximal heart rate and systolic blood pressure in subjects with DS with values obtained for controls at the same exercise time. P < .05 was considered to indicate statistical significance.

slanting palpebral fissures, flat nasal bridge, epicanthal folds, short nose, small dysmorphic ears, short neck with pterygium colli, and brachydactyly. Anthropometric studies showed that 18 (43%) of 42 subjects were obese (weight, 85th percentile); 10 (24%) of 42 had short stature (height, 3rd percentile); and 17 (40%) of 42 had microcephaly (head circumference, 3rd percentile). One subject also had neurosensorial deafness. Only 2 subjects (aged 5 and 6 years) had mild hypotonia.

NEUROPSYCHOLOGICAL AND CARDIORESPIRATORY EVALUATIONS

Thirty-five subjects completed the neuropsychological assessment; the other 7 subjects were unable to do so owing to their scant collaboration. According to the Leiter International Performance Scale,4 20 subjects had mild mental retardation (IQ, 60-69); 8, moderate (IQ, 41-59); and 3, severe (IQ, <40). Two of 35 subjects had normal IQs (80-120), and 2 had borderline IQs (70-79).

Most subjects (37 of 42) achieved (maximal) exercise testing during testing. None of the subjects had symptoms, such as syncope, chest pain, dyspnea, or arrhythmias during testing. Subjects tested had low exercise tolerance (mean ± SD, 61% ± 12%). Only 3 of 37 subjects had exercise tolerance within reference range (80%-100% of normal values6); 27 had reduced tolerance (50%-80%); and 7 had low tolerance (≤50%). The maximal heart rate was 91% ± 4%, and maximal systolic blood pressure was 89% ± 8%. In the Figure, the heart rate was higher in subjects with DS during all exercise.

Subjects with DS had a significantly higher mean ± SD maximal heart rate than controls as determined at the same exercise time (176 ± 10 beats per minute vs 156 ± 11 beats per minute, respectively) (P < .001). Thirty-one of 37 subjects achieved reasonable or good compliance during exercise testing: 3 achieved excellent compliance (score, 5); 16, good compliance (score 4); 12, reasonable compliance (score, 3); 5, poor compliance (score, 2); and 1, low compliance (score, 1). Compliance scores correlated significantly with exercise time (r = 0.62; P < .001) and age (r = 0.44; P < .01) but not with IQ.

Only 18 subjects completed lung function testing. Findings for 10 subjects were normal, and findings for 8 subjects revealed reduced forced vital capacity for those with less compliance and reduced IQ. The main difficulty was for complete aspiration to occur. Parents’
answers to the questionnaire disclosed that only 5 (11%) of the children with DS engaged in regular physical activity.

COMMENT

Numerous studies have focused on people with DS who undergo surgery for congenital heart disease, whereas their counterparts without heart disease remain largely unstudied. Although people with DS without congenital heart disease unquestionably have a better life expectancy, they are at greater risk of the common cardiovascular factors (sedentariness, obesity, and hypertension) than those without DS. As many as 43% of the subjects we studied were obese, and almost 90% were sedentary. These findings provide evidence that until recently most parents of children with DS were poorly informed about their child's fitness level and ability to engage in physical activity. Currently, a growing number of children with DS request permission to participate in sports so that they too can join in the everyday activities that their healthy peers enjoy. Many participate in the Special Olympics. Italian law requires that those wishing to engage in regular sports activities must obtain a medical certificate. In this study, we assessed children with DS with the test protocol that we routinely use for sick children.

Our results show that compared with control subjects of similar age, children with DS without congenital heart disease have a reduced tolerance for exercise, higher heart rates, and higher arterial blood pressure. To obtain more detailed information on tests of a shorter duration, we compared heart rate and systolic blood pressure of subjects with DS with the heart rate and blood pressure of control subjects at the same exercise times. The statistical analysis confirmed that our subjects had significantly higher mean maximal heart rates than controls (P<.001). One reason for the increased heart rate, given that only 11% of our subjects engaged in regular physical activity, could be their lower level of training. A concurrent cause could be obesity. Another explanation might be the novelty of undergoing a previously unknown test. Although the heart rate of children increases with excitement or fear, this should normalize during testing. Our subjects exhibited scarce ability to adapt (Figure).

Our findings imply that collaboration during testing depends more on the individual's mechanical ability to do the technical action required, possibly in a calm atmosphere (relaxed surroundings, operators capable of explaining the test on the subject's level of understanding, and eventual recourse to “test-retest”), than on their IQ. Accordingly, we found no significant correlation between IQs and the degree of collaboration.

The absence of exercise-induced symptoms (syncope, chest pain, or dyspnea) and arrhythmias confirmed our subjects' satisfactory cardiovascular conditions. Because many of our subjects were overweight and had a mild hypertensive response, monitoring these cardiovascular risk factors seems important in these subjects because of the risk for early atherosclerosis. Our subjects' low forced vital capacity during lung function testing probably depended on their difficulty in participating in an active test and partly because they lacked exercise experience (running on treadmill).

In conclusion, children with DS without congenital heart disease can usefully undergo cardiorespiratory tests of functional capacity. Further clinical investigations should be done to assess their neuromuscular development. This proper and complete assessment allows the physician to gauge these subjects' fitness levels to determine their capacity for participating in sports activities, thus generating a “virtuous” circle: sport improves coordination, reduces anxiety, enhances athletic performance, and ultimately leads to a better quality of life.

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