Institute of Experimental Morphology, Pathology and Anthropology with Museum
Bulgarian Anatomical Society

Acta morphologica et anthropologica, 23
Sofia ● 2016

Down Syndrome – Anthropological Point of View: Review

A. Dimitrova

Institute of Experimental Morphology, Pathology and Anthropology with Museum, Bulgarian Academy of Sciences

In the recent years the interest to the morphological changes in the anthropological status in a number of diseases and their use as an additional diagnostic tool considerably increased. The disease with characteristic morphological manifestation is Down syndrome (DS) – most common genetic abnormalities. The individuals with DS are characterized with: short stature, microcephalia or brachicephalia, short extremities, wide hands and short fingers, clinodactyly, syndactyly, etc. The youths with DS demonstrate a great prevalence of overweight and obesity during the puberty. Nowadays anthropological characteristics of children with DS are subject of research in many countries. Knowledge of body composition in children with DS is of interest to clinicians and scientists because components of the body often provide more useful information than the measurements of weight, height or body mass index. To prepare and use DS specific growth charts is important for diagnosis, because growth in children with DS differ from that of their healthy peers.

Key words: Down syndrome, growth charts, short stature, obesity, anthropometry.

In the recent years the interest in the morphological changes in the anthropological status in a number of diseases and their use as an additional diagnostic tool considerably increased. Such disease characterized with morphological manifestation is Down syndrome.

Trisomy 21 is the most common genetic abnormalities, caused by non-disjunction of gametes in the first meiotic division. It occurs in 1 of 700 live births in all ethnic groups and one of the few aneuploidies compatible with postnatal survival. In Bulgaria each year about 100 children with DS are born. Characteristic manifestations of DS are: learning disability, early-onset Alzheimer disease, childhood leukaemia, congenital heart disease, duodenal stenosis, imperforate anus, Hirschprung disease, muscle hypotonia, immune system deficiencies [10]. The most phenotypic features vary among the patients. The individuals with DS are characterized with following morphological features: short stature, microcephalia or brachicephalia, short extremities, wide hands and short fingers, clinodactyly and syndactyly etc. Cytogenetic studies showed that extra chromosome 21 is transmitted through the maternal chromosome in 95% of cases [3]. It is widely assumed that the DS complex phenotype results from the imbalance of the genes located on D21S55 region [9].

Down syndrome is a disease which has existed since ancient times. Evidence can be found at many paintings from Antiquity. In "Madonna and Child" by A. Mantegna (15th c.); "The Peasant and the Satyr" by J. Jordanes (1635-40); "Woman with child" by Jacob Jordaens (1616); "Lady Cockburn and her Children" (1773) by Joshua Reynolds the subjects appear to show the stigmata of Down syndrome. These children which are illustrated at the above mentioned paintings, show different features typical for trisomy 21: prominent tongue, a small nose, an open mouth and slanted eyes.

The archeological data from different parts of the world indicates about cases of burials of individuals with trisomy 21. During excavations on the island of Santa Rosa, CA, a skeleton of a woman approximately around 5200 BC was found. The skull had the typical signs of the disease: flat face, low and wide nasal apertures. It is assumed that it is the oldest finding of an individual with trisomy 21 detected so far. A detailed description of the skull of an individual with Down syndrome found in Leicestershire, England was published in 1960. The rarity of ancient Down syndrome skeletons was unsurprising because of the smaller populations with a younger age structure and higher infant mortality than in modern times [22].

In 1938 the French doctor Escirol for the first time described a child with trisomy 21. The name of disease comes from the name of the British physician John Langdon Down. In the hospital where he worked, he observed a group of individuals with specific phenotype characteristics and mental retardation. For 10 years of work he was able to introduce its rehabilitation training models [10]. Even then it became clear that good medical care, the adequate training and socialization of the patients have a good impact on their health and quality of life.

Down syndrome is a chromosome disorder, well studied in genetic aspect all over the world. From the anthropological point of view, most of studies from 90 years of 20th century and today have been held in the USA. Nowadays anthropological characteristic of children with DS is a subject of research in many countries and specific growth charts for height and weight have been made [7, 26, 1, 19, 16, 23, 24, 25, 18, 11]. The conclusion which can be drawn is that: the preparation of such charts for each country separately, will provide a more reliable assessment of physical development and health of patients with DS.

Height, weight and head circumference are basic measurements used in medical practice and physical anthropology. During the childhood there are important diagnostic signs for the health status, and any deviation from the standard norms indicates the presence of disease. The human stature is genetically determined and has age, sexual, racial and territorial specificity, and it also could be influenced by the environmental factors. Problems associated with growth are common in children. As low growth can be defined the growth with 2 or more standard deviations (SD) below the average rate for age and gender. DS is one of the diseases manifested by variations in basis anthropometric measurements – short stature, microcephaly, low birth weight and risk of overweight and obesity mainly in puberty. Children and adolescents with DS have a set of health, anatomical, physiological and cognitive attributes predisposing them to limitations on their physical activity capacities.

A detailed analysis of the morphological status of children with DS was made in Poland and it determined a tendency to reduce the height and length indexes of the upper limb (arm and forearm lengths). Reports of the measurements of the newborns with DS showed shorter gestation periods (176.5 days), lower birth weights (1.5SD) and birth length reduced by 2SD from the mean of control study [5]. Head circumference showed 1.5 SD from the health controls of the same age. There are no significant sex differences in the patients with DS in this earliest period of physical development [12].

Childhood and adolescence are stages of human life associated with the dynamic growth and development processes in the body. They are the periods of greatest accumulation of muscle, fat and bone mass density (BMD). High levels of BMD are a key determinant for adult skeletal health [20, 21]. The children and adolescent with DS have low BMD and BMDH (BMD/height), compared with non-DS peers [4, 15]. This is the main reason of osteoporotic problems in adulthood in population with DS [2]. The hip region, especially the femoral neck is very important area to be studied, because there is a high risk of bone fracture [14]. Effort to develop physical activity programs, which may enhance bone mass should be considered.

Some authors indicate that the youth with DS demonstrate a great prevalence of overweight and obesity, during the puberty after 10 years of age. In healthy individuals puberty occurs around 10-11 years of age in female and about 12-13 years of age in male and ends in 17th in both sexes. In different investigations of growth and physical development of children with DS found that the puberty begins and ends earlier than their peers. The final height in boys and girls with DS reached 163.4 cm and 151.8 cm respectively [5, 6, 24].

According to some authors there is a relationship between somatotipological characteristics (somatotype) of the patients with DS and some musculoskeletal deformities. Somatotype gets important biometric data that defines the shape and proportionality of human body. It depends on inheritance, the geografical and socio-economical factors. The patients with DS have a great prevalence of endomorph component which determines the presence of overweight and obesity and leads to increased risk of cardiovascular disease, hypertension, diabetes. Therefore annual monitoring of nutritional status in patients with DS is necessary. Endomorphy somatotype is in high correlation with musculoskeletal deformities such as flat foot, forward abdomen and lumbar lordosis, in adolescents with trisomy 21 [8].

Knowledge of body composition in children with DS is of interest to clinicians, and scientists because the components of the body often provide more useful information than the measurements of weight, height or body mass index (BMI). Children with DS have a genetic predisposition to become overweight. Bioelectrical impedance can be a useful technique for body composition analysis in healthy individuals and in those with a number of chronic conditions such as diabetes mellitus, dialysis patients, and patients at risk of obesity. Most studies on body composition of children with DS include height, weight and BMI, but only in few of them is used the bioelectrical impedance analysis (BIA) [13]. Another method, used to study the body composition in healthy individuals and in patients with DS is dual-energy X-ray absorptiometry (DXA). It can be used to more accurately determine adiposity in population with DS. Percentage body fat (PBF) determined by DXA correlated well with PBF determined by BIA in both sexes [17].

In Bulgaria anthropological investigations of individuals with trisomy 21 are very rare and mainly associated with dermatoglyphic features and psycho-physical characteristics [27, 28, 29]. The finger papillar patterns are genetic determinated and very variable in form and size, and may be classified in: loops (L), arches (A) and whorls (W). One of the important quantitative features is the finger ridge count. It consists of the number of ridges, which cut or touch a straight line running from the triradius to the core or center of the pattern. Patients with DS have a great prevalence of loops of the distal phalanges of the fingers, centrally located axial triradius-t and significantly lower finger ridge count on fingers and palmar areas. Normally there are three major flexion creases on the palmar surface of the hand. It may be found in 12 weeks embryos. Abnormal flexion creases can be found in different genetically diseases, like DS. As it is known in 20% of cases of DS there is an additional IV abnormal flexion crease [29].

Morphological investigation of children with mental retardation is known from distant 1972 year [28]. The study included psycho-physical characteristics of children, as 102 of them have a DS. The author established a delay of physical development and stepwise reduction of the height and length of the upper and lower limbs.

Plenty of researches of the anthropological status of the children with DS all over the world and only few in Bulgaria suggest that detailed anthropological characteristics of Bulgarian children and adolescents with DS and specific growth tables and curves need to be developed. This will provide the possibility of monitoring of physical development of individuals with DS and to established deviations from the norms in every age period. This is important for prevention of early occurred complications, related to their physical development.

References

1. **Aburawi, E. H., N. Nagelkerke, A. Deeb, S. Abdulla, Y. M. Abdulrazzaq.** National Growth Charts for United Arab Emirates Children with Down syndrome from birth to 15 years of age. – J. Epidemiol., **25**(1), 2015, 20-29.
2. **Angelopoulou, N., V. Souftas, A. Sakadamis , K. Mandroukas.** Bone mineral density in adults with down syndrome. – Eur. Radiol., **9**, 1999, 648-651.
3. **Antonarakis, S. E., R. Lyle, R. Chrast, H. S. Scott.** Differential gene expression study to explore molecular pathophysiology of Down syndrome. – Brain Res. Rev., **36**, 2001, 265-274.
4. **Baptista, F., A. Varela, L. B. Sardinha.** Bone mineral mass in males and females with and without Down syndrome. – Osteoporos. Int., **16**, 2005, 380-388.
5. **Buday, J.** Growth and physique in Down syndrome children and adults. – Humanbiol. Budapest, 1990, 1-117.
6. **Cannalire, G., M. Bellini, G. Biasucci.** The importance of knowing growth and pubertal development in Down syndrome. – J. Clin. Case Rep., 5:640, 2015, doi: 10.4172/2165-7920.1000640
7. **Cronk, C., A. C. Crocker, S. M. Pueschel, A. M. Shea, E. Zackai, G. Pickens, R. B. Reed.** Growth charts for children with Down syndrome: 1 month to 18 years of age. – Pediatrics, **81**, 1988, 102-110.
8. **Dehghani, L., M. Hashemi, R. Saboonchi, A. Hematfar, A. Roonasi.** Relationship between somatotype and some of musculoskeletal deformities of girl students with Down syndrome. – European Journal of Experimental Biology, **2**(4), 2012, 1209-1213.
9. **Delabar, J. M., D. Theophile, Z. Rahmani, Z. Chettouh, J. L. Blouin, M. Prieur, B. Noel, P. M. Sinet.** Molecular mapping of twenty- four features of Down syndrome on chromosome 21. – Eur. J. Hum. Genet., **1**(2), 1993, 114-124.
10. **Epstein, C. J., J. R. Korenberg, J. Anneren, S. E. Antonarakis, S. Ayme, E. Courchesne, L. B. Epstein, A. Fowler, Y. Groner, J. L. Huret, et al.** Protocols to establish genotype- phenotype correlations in Down syndrome. – Am. J. Hum. Genet., **49**, 1991, 207-235.
11. **Fernandes, A., A. P. Mourato, M. J. Xavier, D. Andrade, C. Fernandes, M. Palha.** Characterisation of the somatic evolution of Portuguese children with Trisomy 21-Preliminary results. – Down syndrome Research and Practice, **6**(3), 2001, 134-138.
12. **Horvath, L., J. Buday.** Birth data of patients with Down syndrome. – Anthrop. Kozl., **30**, 1986, 93-96.
13. **Grammatikopoulou, M. G., A. Manai, M. Tsigga, A. Tsiliqiroqlou-Fachantidou, A. Galli-Tsinopoulou, A. Zacas.** Nutritient intake and anthropometry in children and adolescents with Down syndrome. – Developmental Neurorehabilitation, **11**(4), 2008, 260-267.
14. **Guijarro, M., C. Vabro, B. Paule, M. J. Gonzales, J. A. Riancho.** Bone mass in young adults with Down syndrome. – J. Intellect Disabil., Res., **52**, 2008, 182-189.
15. **Kao, C. H., C. C. Chen, S. T. Wang, S. H. Yeh.** Bone mineral density in children with Down's syndrome detected by dual photon absorptiometry. – Nucl. Med. Commun., **13**, 1992, 773-775.
16. **Lopes, T. S., D. M. Ferreira, R. A. Pereira, G. V. da Veiga, V. M. R. de Marins.** Assessment of

anthropometric indexes of children and adolescents with Down syndrome. – Journal de Pediatria (Rio J), **84**(4), 2008, 350-356.
17. **Loveday, S. J., John M. D. Thompson, E. A. Mitchell.** Bioelectrical impedance for measuring percentage body fat in young persons with Down syndrome: Validation with dual- energy absorptiometry. – Acta Pediatrica, **101**, 2012, pp.e491-e495.
18. **Myrelid, A., J. Gustafsson, B. Ollars, G. Anneren.** Growth charts for Down`s syndrome from birth to 18 years of age. – Arch Dis Child, **87**, 2002, 97-103.
19. **Raja, C. G., C. Parkash, B. V. Bhat, R. K. Ramachandra, B. Rajesh.** Anthropometric measurements in Down's syndrome children during preschool period. – Curr. Pediatr. Res., **11**(1& 2), 2007, 17-20.
20. **Rizzoli, R., J. P. Bonjour.** Determinants of peak bone mass and mechanisms of bone loss. – Osteoporos. Int ., **9** (2), 1999, 17-23.
21. **Rizzoli, R., M. L. Bianchi, M. Garabedian, E. A. McKay, L. A. Moreno.** Maximizing bone mineral mass gain during growth for the prevention of fractures in the adolescents and the elderly. – Bone, **46**, 2010, 294-305.
22. **Starbuck, J. M.** On Anticity of Trisomy 21: Moving towards a quantitative diagnosis of Down syndrome in historic material culture. – Journal of contemporary anthropology, Vol. II, Iss.1, 2011, 21-32.
23. **Styles, M. E., T. J. Cole, J. Dennis, M. A. Preece.** New cross sectional stature, weight, and head circumference references for Down's syndrome in the UK and Republic of Ireland. – Arch. Dis. Child, **87**, 2002, 104-108.
24. **Tuysuz, B., N. T. Goknar, B. Ozturk.** Growth charts of Turkish children with Down syndrome. – An. J. Med. Genet. A., **158A**(11), 2012, 2656-64.
25. **Van Gameren-Oosterom, H. B., P. Van Dommelen, A. M. Oudesluys-Murphy, S. E. Buitendijk, S. van Buuren, J. P. van Wouwe.** Healthy growth in children with Down syndrome. – Plos One, **7**, 2012, e31079.
26. **Zemel, B. S., M. Pipan, V. A . Stalling, W. Hall, K. Schadt, D. S. Freedman, P. Thorpe.** Growth Charts for children with Down syndrome in the United States. – Pediatrics, **136**(5), 2015, e1204-e1211.
27. **Елазарова, Л. Д.** Дерматоглифски проучвания при деца с вродени малформации и техните родители. Автореферат на дис. София, Труд, 1983.
28. **Мутафов, С.** Психо-физически особености на децата с аномалии. София, Медицина и физкултура, 1981.
29. **Торньова-Ранделова, С.** Дерматоглифика при здрави деца и деца със зрителна, слухова и интелектуална недостатъчност. Автореферат на дис. труд. София, 1986.