REVIEW

Bali Medical Journal (*Bali Med J*) 2020, Volume 9, Number 1: 51-54
P-ISSN.2089-1180, E-ISSN.2302-2914

Published by DiscoverSys

Physiological and morphological peculiarities of children with Down's syndrome: A brief review

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ABSTRACT

Background: There are several approaches to solve the problems of many pathological processes' correction, such as clinical pathology and experiment in modern science. At the same time, genetically-conditioned diseases usually demand researchers' attention and generalization of earlier information for making the correction foundation of their manifestations in the future. Down's syndrome is considered to be one. Our objective was to review existing literature regarding physiological and morphological peculiarities of children with Down's syndrome.

Methods: A total of 30 relevant literature were reviewed regarding physiological and morphological peculiarities of children with Down's syndrome. The data collection for eligible articles were conducted from 1989-2017. Different database and manual search methods were used to find the topic-related articles.

Results: Abnormalities of heart, gastrointestinal tract and the system of urinary excretion are considered as the underlying congenital abnormalities for children with Down's syndrome influencing their lifespan. People with Down's syndrome have difficulties in their cognitive activity, such as lag in development, speaking, and reading. Limitation existence of ideas and scarcity of conclusions make studying different school subjects impossible for children with Down's syndrome. They need more time to master new skills and remembering new material. On the other hand, children with Down's syndrome possess positive personal qualities: they are tender, friendly and even-tempered.

Conclusion: Down's syndrome is considered as one of the most common genetic diseases occurred newborns. Children with Down's syndrome have several physical abnormalities. It is important to understand children with Down's syndrome as a whole, as they can be put into a well-adjusted system of rehabilitation.

Keywords: Down's syndrome, children, trisomy 21, morphological and functional disorders.

Cite This Article: Makhov, A.S., Medvedev, I.N. 2020. Physiological and morphological peculiarities of children with Down's syndrome: A brief review. *Bali Medical Journal* 9(1): 51-54. DOI:10.15562/bmj.v9i1.1099

INTRODUCTION

Despite the serious successes of medicine, which were reached in treating socially-significant diseases, conducting of clinical and experimental research, appreciable pathological aggravation attracting the attention of different specialists, remains in modern society.[1-7] Many researchers are inclined to connect the given circumstance with the existence of genetic mechanisms in the development of most diseases, and their etiological treatment will be possible only in the future.[8-10]

It is more challenging to deal with the pathology of diseases, which has only genetic mechanisms in its basis and affects many organs' systems in an individual at once. Down's syndrome is considered to be one of the most widespread genetic diseases of an individual caused by trisomy of the 21st chromosome.[11] This disease was first described by a British scientist John Langdon Down in his scientific work in 1866, whereas the aetiology was determined only in 1959.[12]

It is precisely established that a child with Down's syndrome has little dependence on its parents' way of life, their age and health, bad habits, nutrition, social status, education, nationality, race. There are some rather typical changes in a child's body at the existence of the given syndrome. Modern medicine studies these changes in the course of fundamental and applied investigations.[13,14] It provided an accumulation of scientific knowledge in the given syndrome, of which gradually widens possibilities of its manifestations' correction.[15,16] Frequent occurrence of children with Down's syndrome and also the complexity of somatic aspect and psychical disturbances generalize the most recent information about the current urgent state. This article aims to conclude and present the most critical mechanisms of development and morpho-functional manifestations of Down's syndrome briefly.

Aetiology and pathogenesis of Down's syndrome

The frequency of children born with Down's syndrome is one in 700-800 babies.[17] The frequency of children with Down's syndrome decreased to 1 in 1200-1300 newborn children in the result of antenatal diagnostics and conduction of timely abortion. Mother's age is considered as the main factor influencing the chance of conception of a child with Down's syndrome. If the mother's age between 20 to 24 years old, the possibility is 1 in 1562, before

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Received: 2018-02-28
Accepted: 2020-01-03
Published: 2020-04-01

30 years old – 1 in 1000, from 35 to 39 years old – 1 in 214. If the mother's age is older than 45 years, the possibility is 1 in 19.[17] At the same time, nearly 80% of all born children with this syndrome fall on mother's age before 35. It is conditioned by an increase in pregnancy frequency at this age. Father's age has almost no influence on the risk of a child's born with Down's syndrome.[18] There is an opinion that another risk factor is solar activity. Increase the intensity of ultraviolet radiation connected with cyclic processes on the sun, may influence the processes of an ovum's ripening and increase the risk of chromosomes' nondisjunction appearance during meiosis. There is information that children with Down's syndrome can more often be born in marriages between immediate relatives. It is connected with the fact that immediate relatives are carriers of the same pathologies. That is why, if such a couple had defects of the 21st chromosome, then the possibility of birth of a child with Down's syndrome would increase significantly .[19]

Some of the following mechanisms of Down's syndrome appearance are noted nowadays. Disturbances of meiosis which lead to trisomy of the 21st chromosome, develop in 95% because of the disturbance of ovum's formation and only in 5% - in the result of spermatogenesis disturbance. Most meiosis disturbances in mothers take place in meiosis phase I. The average mothers' age is 32 years. Disturbances in meiosis phase II on mothers take place approximately in 20% of cases. Besides, the disturbances of spermatogenesis leading to nondisjunction of the 21st chromosome may also take place in meiosis phase II.[17] At mosaicism nondisjunction takes place at early stages; thus the disturbance touches only some organs and tissues. Given the variant of Down's syndrome development is called "mosaic Down's syndrome" (46, XX/47, XX, 21). Mosaicism can appear as a result of postzygotic (mitotic) nondisjunction of the normal zygote or postzygotic loss of the 21st chromosome by trisomic zygote.[20] It is cleared up that all these translocations originally came from the maternal origin. At that, the average age of such mothers is 29.2 years old.[18] So, the causes of the appearance of the children with Down's syndrome are mainly revealed. However, the effective treatment of this state will be possible only in the future.

Morpho-functional peculiarities of children with Down's syndrome

Abnormalities of heart, gastrointestinal tract, and the urinary tract system are considered as the basic congenital abnormalities for children with Down's syndrome, which later affect their lifespan. The main cause of early mortality of children with Down's syndrome is congenital heart diseases, which are found nearly in 50% of cases. These are the common heart defects found: a defect of the interventricular septum, an opening of the arterial duct, defect of the interatrial septum, and Fallot's tetrad. Their existence is aggravated by common morpho-functional immaturity of such newborn babies.[13] Abnormalities of gastrointestinal tract development are met in 4-10% of children with Down's syndrome and can remain clinically asymptomatic for some months. Research showed that the common gastrointestinal tract problems are: esophageal atresia (0.3-0.8%), pyloric stenosis (0.3%), stenosis/atresia of duodenum (1-5%), Hirschsprung's disease (1-3%), and anal stenosis/atresia (1-4%). Frequent constipation in children with trisomy of the 21st chromosome is explained by the existence of muscle hypotonia and flaccid intestinal motility.[21] The frequency of congenital abnormalities of the urinary tract system in children with Down's syndrome (hydronephrosis, pyelectasis. and renal hypoplasia/aplasia) reaches 4%.[22]

Furthermore, thyroid gland diseases can commonly be found in Down's syndrome. These diseases are registered in these children approximately in 28-40% of cases. Their quantity increases with ageing and reaches 54% in 2013. Foreign research noted that there is a quite high prevalence of subclinical hypothyroidism at this state. Congenital autoimmune thyroiditis (Hashimoto) is observed rather seldom.[23] Pancreatic diabetes is found approximately 1% of children with Down's syndrome. The course of the disease has no specific character.[13] Defects in the musculoskeletal system of children with Down's syndrome are characterized by weakness of ligamentous apparatus, hypermobility of joints, and muscle hypotonia. Dysplasia of connective tissue lies in their basis, caused by the abnormal structure of collagen fibres. In addition, nearly half of such children have scoliosis. Atlantoaxial instability is also a manifestation of this pathology, which is met in 10-15% of cases of Down's syndrome.[24] Nearly 5-8% of children have the pathology of hip joints, such as hip subluxation. Children with Down's syndrome are also characterized by the frequent occurrence of slipped femoral whirlbone epiphysis, which is connected to obesity and hypothyroidism. Prevalence of patella's luxation and subluxation was found in 20% of children. In most cases, the children have moderate subluxation with painless restriction of joint's movements. Platypodia is typical for nearly half of the children with Down's syndrome and its severe forms demanding operative intervention, are rather frequent.[24]

Published by DiscoverSys | *Bali Med J* 2020; 9(1): 51-54 | doi: 10.15562/bmj.v9i1.1099

Anatomical craniofacial anomalies which are typical for children with Down's syndrome, lead to the development of different diseases of ENT-organs. Narrow nasal ducts and small oral cavity at trisomy of the 21st chromosome can disturb nasal breathing. The existence of macroglossia and macrogenia also disturb the child's breathing as the child breathe mostly through the mouth. Thus, it increases the frequency of respiratory infections and periodontitis. Children with Down's syndrome are noted to have a change in dental structure: little crow with broad and short roots, promoting early loss of teeth. Nearly half of the children with Down's syndrome also have partial anodontia.[13] Frequent otitis arising against this background leads to the development of hearing loss in 38-78% of children with Down's syndrome. This frequent otitis was due to short and horizontal disposed Eustachian tube, promoting the accumulation of liquid in the middle ear.

Most children with Down's syndrome have disturbances from the side of eyesight organs. They often have strabismus (20-47%), nystagmus (11-29%), congenital and acquired cataract (4-15%), blepharitis (7-41%), disturbance of refraction (43-70%), and glaucoma (0.7%). Stenosis of nasolacrimal canals is often observed at trisomy of the 21st chromosome.[17] Oncological diseases are rather widespread among children with Down's syndrome. Many newborn babies with trisomy of the 21st chromosome are observed to have changes in hemopoiesis regulation leading to such disturbances such as polycythemia, thrombocytopenia, thrombocytosis, and others. As a result, the risk of leukaemia development in them is in 18 times higher than in healthy children. More than 10% of newborn babies with Down's syndrome have a leukemoid reaction in their blood analyses. This reaction is considered to be connected with the additional 21st chromosome.[13] To conclude, children with Down's syndrome are characterized by some physical disturbances. It should be taken into account as their decompensation may lead to an untimely death.

Psychical peculiarities of development of children with Down's syndrome

All children with Down's syndrome have difficulties in their cognitive ability. They significantly lag in development and begin to speak and read much later.[25] Lagging in speech development is caused by several factors, such as gaps in mastering the grammatical constructions, the ability to remember new words and grammatical rules is lowered, and difficulties in understanding the tasks.[26] Thus, children with Down's syndrome have fewer vocabulary knowledge. Problems in speech development, leading to a lack of participation in communication for such children. Parents or grown-ups tend to ask them questions and the children with Down' syndrome need time to answer questions. They need time to comprehend new words and structures of sentences, and thus sometimes grown-ups feel like the children are lagging in speech.[27]

Children with Down's syndrome have significant inability to develop reasoning and alignment of proofs. They hardly transfer skills and knowledge from one situation to another. Abstract notions are unavailable for their understanding as solving practical problems is very difficult for them. Limitation of ideas and scarcity of conclusions lying based on intellectual activity, make studying different school subjects impossible for children with Down's syndrome. The existence of hypomnesia in them aggravates it. They need more time to master new skills and new material. They are also characterized by instability of active attention, increased fatigability and exhaustibility. Down's syndrome children take everything just visually; furthermore, they cannot correlate parts of drawing with each other and unite them into a whole picture.[26] However, there is no evidence in terms of children with Down's syndrome imaginations.

The ability characterizes the behavior of children with Down's syndrome to subordination, good nature, and sometimes tenderness. Besides, children with Down's syndrome are noted to have preservation of elementary emotions, such as tender and affectionate. Some of them show positive emotions to all the grown-ups, communicate with them; some of them are predominate with those who they are used to communicate. Such children cannot estimate the results of their activity and the emotion of pleasure usually accompanies the completion of the task, which at that may be fulfilled incorrectly. They also feel fear, joy, and sadness. However, the depth of emotional reactions does not correspond to their causes. They are not often expressed brightly enough, although there may be intense feelings caused by insignificant grounds.[28] These children are mostly characterized by their suggestibility, imitativeness to actions, and deeds to others. Epileptoid traits of character are observed in just some of them, which are egotism and excessive tidiness. However, most children with Down's syndrome are tender, friendly and even-tempered.[25] In conclusion, children with Down's syndrome are characterized by some psychical disturbances which should be taken into account in the course of correcting the state.

CONCLUSION

Down's syndrome is considered as one of the most common genetic diseases occurred newborns. Children with Down's syndrome have several physical abnormalities, such as abnormalities of the heart, gastrointestinal tract, and urinary tract. In addition, they have late cognitive ability development; thus, they need more time to comprehend and learn words. As for the personality, generally, they are tender, friendly, and even-tempered. To conclude, it is important to understand children with Down's syndrome as a whole, as they can be put into a well-adjusted system of rehabilitation.

CONFLICT OF INTEREST

The author declares that there is no conflict of interest regarding all aspect of the study.

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