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Congenital Anomalies: Public Health Interventions to Ensure its Prevention and Expansion of Care to the Patients

Konjenital Anomaliler: Halk Sağlığı İçin Müdahaleleri Sağlamak, Olumsuzlukları Önlemek ve Hasta Bakımının Geliştirilmesi

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ABSTRACT

Congenital anomalies can be defined as structural or functional anomalies, including metabolic / biochemical disorders, which are present at the time of birth. Congenital anomalies has been recognized as a major public health concern, owing to its universal distribution, associated long-term disability; social stigma; emotional / psychological stress for the family members; increased medical expenditure; and burden on the health care delivery system & societies. To prevent the occurrence of congenital anomalies, due attention should be given to establishment of appropriate surveillance systems to record cases from both community and hospital settings; strengthening of public health system; promoting research to explore the etiological factors and diagnosis/prevention strategies; fostering international cooperation; and discouraging the practice of consanguineous marriage / conception at an advanced age / further reproduction after birth of a malformed child. To conclude, there is an indispensable need to formulate a comprehensive policy, that should be well-supported by an efficient surveillance system, dedicated health care professionals and involvement of all stakeholders.

Key words: Congenital anomalies, Rubella, Global, Public health

ÖZET

Konjenital anomaliler yapısal veya fonksiyonel olarak tanımlanır ve doğuştan ortaya çıkan metabolik/biyokimysal bozuklukları içerir. Evrensel bir problem olan konjenital anomaliler; uzun süreli bozukluklarla ilişkili olmaları; sosyal stigma, ailelerin duygusal / fizyolojik stres yaşamaları, artan medikal maliyetlerle birlikte sosyal sağlık sistemlerine yük oluşturmaları nedeniyle önemli bir halk sorunu olarak tanımlanmaktadır. Konjenital anomalilerin ortaya çıkmasından korunmak için; toplumdaki ve hastanedeki vakaların kaydedilmesi amacıyla gözetim sistemi kurulmalı, halk sağlığı sistemi güçlendirilmeli, etyolojik faktörlerin araştırılması, teşhis ve korunma stratejilerinin geliştirilmesi için teşvik verilmeli, uluslararası işbirliği yapılmalı ve akraba evliliği/ ilerlemiş yaşlarda gebe kalma/ malformasyona sahip çocuklu ailelerin yeniden çocuk dünyaya getirmeleri konusunda uyarılarda bulunmak gerekmektedir. Sonuç; sağlık profesyonelleri ve gönüllülerin katılımıyla etkin bir gözetim sistemi tarafından desteklenen kapsamlı bir politika sistemi formüle edilmelidir.

Anahtar kelimeler: Konjenital anomali, rubella, global, halk sağlığı

INTRODUCTION

Magnitude & impact on global health of congenital anomalies

Congenital anomalies can be defined as structural or functional anomalies, including metabolic / biochemical disorders, which are present at the time of birth & recognized at an early age[1]. Globally, the recent estimates have indicated that almost 1 in every 33 infants suffer from a congenital anomaly[2]. Furthermore, in excess of 0.25 million newborns succumb to death within the first 28 days of their life every year from congenital anomalies[2]. Congenital anomalies has been recognized as a major public health concern, owing to its universal distribution, associated long-term disability; impact on the quality of life of the child/family members; social stigma; emotional / psychological stress for the family members; increased medical expenditure (viz. direct / indirect medical cost or rehabilitation expenses); and burden on the health care delivery system & societies[2,3].

Potential risk factors

The etiology of most congenital anomalies is poorly understood, and for almost half of them no specific cause can be attributed[2]. However, a wide range of risk factors such as advanced maternal age; ethnicity (viz. Ashkenazi Jews or Finns); consanguineous marriages; poor socio-economic status (enhanced susceptibility to macro/micronutrient malnutrition or infections during antenatal period); chromosomal abnormalities; inborn errors of metabolism; intrauterine infections like syphilis/rubella; exposure to teratogenic drugs or pesticides or alcohol/tobacco; maternal nutritional status (viz. iodine deficiency, folate insufficiency, or diabetes mellitus); irradiation; and exposure to high doses of vitamin-A during early pregnancy (viz. treatment of acne); have been implicated in the causation of congenital anomalies[2,4,5].

Suggested measures

- General measures: Owing to the preventable nature of most of the congenital anomalies, the member states of the World Health Assembly have advocated for the universal promotion of primary prevention strategies; establishment of appropriate surveillance systems to record cases from both community and hospital settings; strengthening of public health system; promoting research to explore the etiological factors and diagnosis/prevention strategies; fostering international cooperation with welfare agencies (viz. World Health Organization / Global Alliance for Vaccines and Immunization / United Nations Children's Fund)[2,6].
- Preventive measures: To prevent the occurrence of congenital anomalies, due attention should be given to the quality of antenatal and post-natal care services; the accessibility and affordability of the services; improving the dietary habits of a women to ensure adequate intake of vitamins and minerals (especially, folic acid and iodine); motivating mothers to either abstain or restrict exposure to drugs/alcohol/tobacco/hazardous environmental substances/radiation; improving immunization coverage against rubella virus; creating awareness among people about congenital anomalies; encouraging use of genetic counseling; discouraging the practice of consanguineous marriage / conception at an advanced age / further reproduction after birth of a malformed child; and in organizing training sessions for health care professionals to have a high index of suspicion for congenital anomalies[3,7].
- Early diagnosis: To promote early detection of congenital anomalies, strategies like preconception screening (viz. to identify persons at risk for specific disorders or at risk for passing a disorder on to their children); antenatal screening (viz. screening for advanced maternal age, Rhesus blood group incompatibility, carrier screening and screening for alcohol, tobacco and other psychoactive substance use); employment of

prenatal diagnostics (viz. using amniocentesis / chorionic villi sampling / ultrasound / estimation of alpha-fetoprotein); and newborn screening (viz. thorough clinical examination and screening for hematological/biochemical disorders); should be employed[2,5].

- Therapeutic care & rehabilitation: If a child is still born with a congenital anomaly, the health care delivery system should be well-equipped to provide them with surgical/medical treatment (viz. blood transfusion – thalassemia, etc.) or psycho-social support services at the earliest to improve their prognosis[2,3].

Critical elements

Nevertheless, none of the above mentioned strategy can deliver good results, if there is no national program for the prevention and care of birth defects, tailor-made to the local settings[2]. In order to ensure a holistic improvement in the quality of life of people with congenital anomalies, there is a crucial need to implement cost-effective interventions to improve health status of the children; development of community-based rehabilitation programs for the welfare of people with disabilities; and building partnership with voluntary non-governmental organizations to expand the reach of services[1,2].

CONCLUSION

To conclude, owing to the enormous magnitude and preventable nature of a major proportion of congenital anomalies, there is an indispensable need to formulate a comprehensive policy, that should be well-supported by an efficient surveillance system, dedicated health care professionals and involvement of all stakeholders.

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