PDF version

Short communication

Khaira Boussouf [1,7], Zoubida Zaidi [2,7], Mounira Amrane [3,7], Naima Hammoudi [4], Malika Mebarki [5]
and Sid Ali Amalou [6]

□□□□□□□ □□ □□□□ □□□□□□□ □□□□ □□ □□□□□□□□ □□□□□ □□□□□ □□□□□

□□□□□ □□□ □□□ □□□□□□□ □□□□□ □□□□□□ □□□□□ □□□□□□ □□□□□ □□□□□ □□□□□ □□□□□□ □□□□

□□□□□□□□□□ □□□□□□□□ □□□□□ □□□□□ □□□□□□ □□□□□□ □□□ □□□□□□ □□□ □□□ □□□□□□□ □□□ □□□□ : □□□□□□□ □□ □□□□□ □□□□□□□□ .□□□□□□□□ □□□□ □□□□□ □□ □□□□ □□□□□□□□ □□□□□□□□ □□□□□□ □□□□□ □□ □□□ .□□□□□□ □□ □□□□□□ □□□□□□□□ □□□□□□□□□□ □□□□ □□ □21 □□□□□□ □□□□□ □□□□□ .2013 □□□□□ □□□□□/□□□□□□ □□□ 2009 □□□□□□ □□□□□/□□□□□ □□ □□□□□□□□□□ □□□□□□□□ □□□□□ □□□□□ □□□□ □□□□□□□□ □□□□□□□ □□□□□□□ □□□□□□□□ □□□□ □□□□□□□ □□□□□ □□□□□ □□□□ □□□□□□□□ □□□□□ □□□□□ □□ (0.06% ± 15.4) □□□□ 22 □□ □□□□□□□□ □□□□□□□ .□□□□□□□□ □□□□□□□□ □□ □□□□□□ □□□□ □□□□□□□ □□□□ □□□□□ □□ □□□□ 143 □□□□□ □□□□□□□ □□□□□□□□□ □□□□□□□□□ □□□□□□ □□□□□□ 770 □□□□□□ □□ (2.2% ± 10.6) □□□□ 88 □□□ □□□□□□ □□ □□□□□□ □□□□ □□□□ □□□□□□□□□ □□□□□□□□□□ □□ □□□□□□ □□□□□□□ □□ □□□□□□□□ □□□□□ □□□□□ □□ □□□□□□ 68) □□□□□□ 75 □□□□□ □□□□□ 110 □□□ □□□ .□□□□ □□□□□□□ □□ □□□□□□ □□□□□ □□□□□□ □□□□□□□ □□□□ □□□□□□ .□□□□□□ □□□□□ □□□□□□ □□ (% 32) □□□□ 35□ □□□□□□ □□□□□ □□□□□□ □□ (% □□□ □□□ □□ □□□□□□□□ .□□□□□□□ □□□□□□ □□□□□□ □□ □□□□□□ □□□□□□□□ □□□□□ □□□□□□□□ □□ □□□□□□□□ □□□□□ □□□□□□□□ □□□□□ □□□□□□ □□□□ □□□□□□□□ □□□□□ □□□□□ □□□□□ □□□□□□□ .□□□□□□ □□□□□□□ □□ □□□□ □□□□□□ □□□□□□□□ □□□□ □□□□□ □□ □□□□ □□□□□□□ □□□□

ABSTRACT This study aimed to describe and evaluate the type, frequency and patterns of congenital heart diseases (CHDs) in patients with Down Syndrome (DS) in Sétif, Algeria. Down Syndrome, or trisomy 21, is the most common genetic disorder in the world. Data were collected and followed from January 2009 to December 2013. Parental consanguinity documenting pedigree analyzing, chromosome analysis and clinical examination were carried out for all cases. Results have shown that 22 (15.4%; ± 0.06) of the total 143 known cases of DS from DS centres have CHDs and 88 (10.6%; ± 2.2) of the total 770 patients with CHDs

collected from public departments at the child and maternity teaching hospital, Sétif, have DS. Among the 110 cases, 75 (68%) have single cardiac abnormalities and 35 (32%) have multiple cardiac abnormalities. The most frequent CHDs were Atrioventricular Septal Defect (AVSD). In conclusion, our study will be helpful to demonstrate the current status of DS and to identify the distribution of CHD in patients with DS in Sétif, Algeria, for further study.

Étude des cardiopathies congénitales chez des patients atteints du syndrome de Down en Algérie

RÉSUMÉ La présente étude avait pour objectif de décrire et d'évaluer le type, la fréquence et les caractéristiques des cardiopathies congénitales chez des patients atteints du syndrome de Down à Sétif (Algérie). Le syndrome de Down, ou la trisomie 21, est le trouble génétique le plus courant au monde. Des données ont été collectées et suivies entre janvier 2009 et décembre 2013. L'étude de la consanguinité des parents incluant l'analyse de l'arbre généalogique, une analyse chromosomique et un examen clinique, a été réalisée pour tous les cas. Les résultats ont montré que sur 143 cas connus de syndrome de Down provenant de centres spécialisés dans la prise en charge de ces patients, 22 (15,4 %; ± 0,06) souffraient d'une cardiopathie congénitale et que sur 770 patients atteints d'une cardiopathie congénitale venant des départements publics des hôpitaux universitaires de soins maternels et infantiles à Sétif, 88 (10,6 %; ± 2,2) étaient atteints du syndrome de Down. Parmi ces 110 cas, 75 (68 %) souffraient d'une seule anomalie cardiaque tandis que 35 (32 %) avaient plusieurs anomalies cardiaques. La cardiopathie congénitale la plus courante était la communication auriculo-ventriculaire. Pour conclure, notre étude sera utile pour mettre en évidence la situation actuelle du syndrome de Down ainsi que pour identifier la répartition des cardiopathies congénitales à Sétif (Algérie) en vue d'études ultérieures sur le sujet.

[1]Department of Cardiology, [2]Department of Epidemiology, [3]Department of Biochemistry, [4]Depa rtment of Cardiology, University Hospital of Ben Aknoun, Algiers, Algeria.
[5]

Department of Paediatrics, University Hospital of Sétif, Sétif, Algeria (Correspondence to: Khaira Boussouf: boussouf.kh@gmail.com).
[6]

Private Cardiology Clinic, Sétif, Algeria.
[7]

Genetics Laboratory of Cardiovascular Diseases, Sétif, Algeria.

Received: 14/09/16; accepted: 21/12/16

Introduction

Congenital heart diseases (CHDs) are a leading cause of birth defects (1,2). Down syndrome, or trisomy 21, is a chromosomal disorder which is often associated with morphological and structural defects. People with Down syndrome are clinically diagnosed by the presence of a characteristic phenotype. The clinical diagnosis is confirmed by chromosome analysis (3). The risk of Down syndrome increases with increasing age of the mother (4).

Among all cases of CHDs, 4–10% have Down syndrome and 40–50% of people with Down syndrome have CHDs (5). They are the most common cause of death in people with Down syndrome in the first 2 years of life (6–8). Therefore, echocardiography is recommended for early detection of CHDs, which may help to prevent many complications. Single heart defects are usually found in people with Down syndrome but multiple defects are also found. In the United States of America and Europe, atrioventricular septal defect is reportedly the most common CHD associated with Down syndrome (9–13). In Asian communities, ventricular septal defect is the most common defect (14,15) whereas in Latin America, atrial septal defect is reportedly the most common defect (11,16).

In recent decades, there has been a substantial increase in the life expectancy of children with Down syndrome in general (17). This increase in life expectancy is mainly due to the successful early surgical treatment of CHD in children with Down syndrome (9,17-20).

There have been no previous studies about CHDs in patients with Down syndrome in Algeria. The aim of this study, therefore, was to determine the type and distribution of CHDs in young patients with Down syndrome in Sétif, eastern Algeria.

Methods

Study design, setting and participants

This case series study examined the type, frequency and patterns of CHDs in young people (

Our study population consisted of all Down syndrome patients with CHDs in Sétif. Patients were drawn from the 6 Down syndrome centres and the university hospital. Primary care physicians in the other hospitals, health centres and primary health care centres have not been trained to treat Down syndrome patients with CHDs so these patient are all referred to these 2 places

The recruitment period spanned 18 months (January 2009 to June 2010) in order to have a large representative sample. Patients were followed until December 2013. Eligible patients were Sétif residents aged

All Down syndrome patients **Data collection**

All potential cases underwent full clinical assessment including phenotypic features that suggested Down syndrome, such as hypotonia, brachycephaly, small low-set ears, upslanting creases and a gap between the first and second toes. Those with Down syndrome and CHDs, and the Down syndrome patients without CHDs from the centres who agreed to participate had a detailed medical history taken of age, sex, consanguinity and family history with special emphasis on maternal and paternal ages. The diagnosis of Down syndrome was confirmed by chromosome analysis and all patients were examined by plain chest X-ray, electrocardiogram and ultrasound of the heart (2-D echocardiography). The echocardiography examination was performed at the cardiology department using a GE Vivid 3 ultrasound machine.

Statistical analysis

SPSS, version 16 was used for analysis. Descriptive statistics were done and data are presented as means and standard deviations (SD) or proportions.

Ethical considerations

This study was approved by the University Hospital and the Faculty of Medicine of Sétif University. Informed consent was given by the guardians of all the participants; none declined.

Results

Among 143 patients with Down syndrome

Table 1 shows the demographic characteristic of the patients: their ages ranged from 3 months to 20 years, with a mean age of 4,3 (SD 0.5) years , 60 (55%) were male and 13 (12%) had consanguineous parents.

The mean ages of the mothers and fathers of the patients were 36.6 and 43.2 years respectively.

Table 2 compares maternal age and parental consanguinity of Down syndrome patients with

and without CHDs. Maternal age 40–49 years was significantly associated with CHDs in Down syndrome patients (P = 0.03) but first and second degree consanguinity was not (P = 0.12 and 0.1 respectively).

Table 3 show the types of CHD among the Down syndrome patients with CHDs. Of the 110 patients, 75 (68%) had a single cardiac abnormality, while 35 (32%) had multiple cardiac abnormalities. The most common CHDs were atrioventricular septal defect isolated (30%) or combined with other cardiac abnormalities (44%), ventricular septal defect (17%), atrial septal defect (7%) and patent ductus arteriosus (6%).

Table 4 shows the distribution of atrioventricular septal defect and ventricular septal defect in the Down syndrome patients by sex. No significant difference by sex was found for either atrioventricular septal defect or ventricular septal defect (P > 0.05).

Discussion

Our study confirms some data reported by other studies on CHDs in people with Down syndrome (13,21). The frequency and distribution of CHDs in Down syndrome varies with geographical region (1). Ours is the first study of CHDs in patients with Down syndrome in a state of Algeria. In our study, the prevalence of CHDs was 15.4% among the Down syndrome patients aged between 5 and 20 years at the Down syndrome centres. This may be because more deaths occurred in patients with Down syndrome and CHDs in the absence of early surgery. The frequency of CHDs in Down syndrome patients observed in our study is less than that reported in Korea (57%), Sudan (43%) and Oman (57%) (16,22,23). However, these studies included infants with a mean age of 3 months versus 52 months in our study. In the hospital sample, among 770 patients with CHDs aged between 3 and 48 months, 11.4% had Down syndrome, which is similar to the prevalence reported in previous studies (4–10%) (1,5,21).

Consanguineous marriage is common in the Middle East and Arab countries, especially in small and rural areas. In the current study, the consanguinity rate among the parents of the Down syndrome patients with CHDs was 12%. This differs from the rate reported in Saudi Arabia (57.8%) (24) and other countries. (25–29).

We found a statistically significant relationship between maternal age 40–49 years and Down syndrome with CHDs; however there was no significant difference with mothers ≤ 39 years. Our data differ from those reported in Korean and Lebanese studies; they found that mothers ≥ 35

years old were more likely to give birth Down syndrome child with a CHD than infants with Down syndrome born to mothers who were

The most common CHDs in our study were atrioventricular septal defect isolated or combined with other cardiac abnormalities, ventricular septal defect, atrial septal defect and patent ductus arteriosus. This is similar to findings of studies in Europe and the United States of America (12,13). However, atrial septal defect was the most common defect reported as in a Korean study (30.5%) (16). In Saudi Arabia, ventricular septal defect was the most common defect (14), while in Singapore patent foramen ovale predominated (30).

In the absence of newborn screening in Algeria, we could only draw patients from Down syndrome centres and the department of paediatrics of the university hospital. So our patients may not be representative of all Down syndrome patients and the prevalence found is only therefore an approximate figure.

Conclusion

This is the first study to document the types, distribution and frequency of CHDs in Algerian patients with Down syndrome. The most frequent CHD diagnosed was atrioventricular septal defect. High maternal age appears to be a risk factor for CHDs in Down syndrome patients in the Algeria population. The characteristics of CHDs in Down syndrome patients from Sétif are similar to those reported worldwide.

Funding: None.

Competing interests: None declared.

References

1. Hoffman JIe. The global burden of congenital heart disease. Cardiovasc J Afr. 2013;24(4):141–5.
2. Bruneau BG. The developmental genetics of congenital heart disease. Nature. 2008;451(7181):943–8.
3. Jung Min Ko. Genetic Syndromes associated with Congenital Heart Disease. Korean Circ J. 2015;45(5):357–61.
4. Ghassan C, Chokor I, Fakhouri H, Hage G, Saliba Z, El-Rassi I. Congenital heart disease, maternal age. and parental consanguinity in children with Down's syndrome. J Med Liban. 2007;55(3):133–7.

5. Greenwood RD, Nadas AS. The clinical course of cardiac disease in Down's syndrome. Pediatrics. 1976;58:893–7.

6. Weijerman ME, van Furth AM, Vonk Noordegraaf A, van Wouwe JP, Broers CJ, Gemke RJ. Prevalence, neonatal characteristics, and first-year mortality of Down syndrome: a national study. J Pediatr. 2008 January;152(1):15–9.

7. Ahmadi A, Koorosh E, Etemad K, Khaledifar A. Risk factors for heart failure in a cohort of patients with newly diagnosed myocardial infarction: a matched, case-control study in Iran. Epidemiol Health. 2016;38:e2016019.

8. Ahmadi A, Soori H, Mehrabi Y, Etemad K, Samavat T, Khaledifar A. Incidence of acute myocardial infarction in Islamic Republic of Iran: a study using national registry data in 2012. East Mediterr Health J. 2015;21(1):5-12.

9. Nisli K, Oner N, Candan S, Kayserili H, Tansel T, Tireli E, et al. Congenital heart disease in children with Down's syndrome: Turkish experience of 13 years. Acta Cardiol. 2008;63:585–9.

10. Weijerman ME, van Furth AM, van der Mooren MD, van Weissenbruch MM, Rammeloo L, Broers CJ. Prevalence of congenital heart defects and persistent pulmonary hypertension of the neonate with Down syndrome. Eur J Pediatr. 2010 Oct;169(10):1195–9.

11. Vilas Boas LT, Albernaz EP, Costa RG. Prevalence of congenital heart defects in patients with Down's syndrome in the municipality of Pelotas, Brazil. J Pediatr (Rio J). 2009;85(5):403-7.

12. Freeman SB, Bean LH, Allen EG, Tinker SW, Locke AE, Druschel C, et al. Ethnicity, sex, and the incidence of congenital heart defects: a report from the National Down Syndrome Project. Genet Med. 2008;10(3):173–80.

13. Stoll C, Dott B, Alembik Y, Roth MP. Associated congenital anomalies among cases with Down syndrome. Eur J Med Genet. 2015;58(12):674–80.

14. Abbag FI. Congenital heart diseases and other major anomalies in patients with Down syndrome. Saudi Med J. 27(2):219-22.

15. Chéhab G, Fakhoury H, Saliba Z, Issa Z, Faour Y, Hammoud D, et al. Cardiopathies congénitales et malformations gastro-intestinales associées [Congenital heart disease associated with gastrointestinal malformations]. J Med Liban. 2007;55(2):70–4.

16. Kim MA, Lee YS, Yee NH, Choi JS, Choi JY, Seo K. Prevalence of congenital heart defects associated with Down syndrome in Korea. J Korean Med Sci. 2014;29(11):1544–9.

17. Holland Bittles A, Bower C, Hussain R, Glasson E. The four ages of Down syndrome. Eur J Public Health (2007) 17 (2): 221-225.

18. Kucik JE, Shin M, Siffel C, Marengo L, Correa A, Congenital Anomaly Multistate Prevalence and Survival Collaborative. Trends in survival among children with Down syndrome in 10 Regions of the United States. Pediatrics. 2013;131(1): e27-36.

19. Leggat S. Childhood heart disease in Australia. Current practices and future needs. Pennant Hills: HeartKids Australia; 2011.

20. Majdalany DS, Burkhart HM, Connolly HM, Abel MD, Dearani JA, Warnes CA, et al. Adults with Down syndrome: safety and long-term outcome of cardiac operation. Congenit Heart Dis. 2010;5(1):38-43.--

21. Ali SKM. Cardiac abnormalities of Sudanese patients with Down's syndrome and their short-term outcome. Cardiovasc J Afr. 2009;20(2):112–5.

22. Elshazeli OH. The spectrum of congenital heart defects in infants with Down's

syndrome, Khartoum, Sudan. J Pediatr Neonatal Care. 2015;2(5):00091.

23. Jaiyesimi O, Baichoo V. Cardiovascular malformations in Omani Arab children with Down's syndrome. Cardiol Young. 2007;17(2):166–71.

24. El-Attar LMA. Congenital heart diseases in Saudi Down syndrome children: frequency and patterns in Almadinah region. Res J Cardiol. 2015;8(1):20-6.

25. Jarallah A. Down's syndrome and the pattern of congenital heart disease in a community with high parental consanguinity. Med Sci Monit. 2009;15(8):CR409-12.

26. Ashraf M, Malla RA, Chowdhary J, Malla MI, Akhter M, Rahman A, et al. Consanguinity and pattern of congenital heart defects in Down syndrome in Kashmir, India. Am J Sci Ind Res. 2010;1(3):573-7.

27. Settin A, Almarsafawy H, Alhussieny A, Dowaidar M. Dysmorphic features, consanguinity and cytogenetic pattern of congenital heart diseases: a pilot study from Mansoura locality, Egypt. Int J Health Sci (Qassim). 2008;2(2):101-11.

28. Abdullah Garjees N, Abdullah Muhsin A. Congenital heart disease in children with Down syndrome in Duhok City. Pak Pediatr J. 2013;37(4):212–6.

29. Ul Haq F, Jalil F, Hashmi S, Jumani MI, Imdad A, Jabeen M, et al. Risk factors predisposing to congenital heart defects. Ann Pediatr Cardiol. 2011;4(2):117–21.

30. Azman BZ, Ankathil R, Siti Mariam I, Suhaida MA, Norhashimah M, Tarmizi AB, et al. Cytogenetic and clinical profile of Down syndrome in Northeast Malaysia. Singapore Med J. 2007 Jun;48(6):550–4.

Thursday 11th of July 2024 08:13:17 AM