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Frequency of Ventricular Septal Defect in Down Syndrome Patients Presenting at a Tertiary Care Hospital

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ABSTRACT

Background: In Down syndrome, ventricular septal defect (VSD) is a typical cardiac abnormality. **Objective:** The estimation of the frequency of diabetes mellitus in Hepatitis C patients is the main objective of this study. **Study Design:** Cross sectional study. **Settings:** The research was done in the Department of Cardiology Shaikh Zayed Hospital, Lahore Pakistan. **Duration:** March 2022 to August 2022. **Methods:** Total sample of participants would be determined by using Raosoftware. The sample size was selected to be n=100 after taking prevalence of 58.3%, 9% margin of error and confidence level at 95 %. The statistical analysis was done by using SPSS version 20. **Results:** The average age of the 100 enrolled individuals who tested positive for hepatitis C RNA was 48.46 9.05 years. With a female to male ratio of 1.9:1, the sample included 66 women and 34 men. There were 55 (55%) people with diabetes out of 100, however 29 (29%) were found accidentally during routine laboratory testing; the other people were either on dietary restrictions or weren't taking their prescriptions as prescribed. The majority of the patients (n=24, or 24%) had HBA1C >7.6 i.e., while another 23 (or 23%) had HBA1C between 6.5-7.0%, and the other 8 (or 8%) had HBA1C between 7.1-7.5%. **Conclusion:** Due to the increasing incidence of diabetes in HCV RNA-positive patients, studies are necessary to develop viable and inexpensive techniques for the monitoring and treatment of diabetes in hepatitis C patients. These strategies must be suitable for use in hepatitis C patients.

Keywords: *Chronic liver disease, Diabetes mellitus, Hepatitis C.*

INTRODUCTION

About one in every 700 newborns is affected with autosomal trisomy (C21), the most widespread type of which is Down syndrome. Protein-coding genes number about 225, with an additional 165–404 non-protein-coding RNAs (ribonucleic acid) acting as regulatory factors make up human C21, also known technically as Has 21.[1,2] Abnormalities in the heart, neurological system, gastrointestinal tract, and reduced sizes of the frontal and temporal cortices, the hippocampus, the cerebellum, and the brain stem are all associated to the uncontrolled synthesis of proteins due to gene triplication. In addition, irregularities in brain connection have been detected.[3,4] Between 1% and 3% of the population is thought to have an intellectual handicap. Up to 28% of intellectual impairments are caused by chromosomal abnormalities (both numerical and structural), with aneuploidies being a common cause.[5] Autosomal trisomy (chromosome 21-C21) is the most common kind of genetic abnormality, with Down syndrome being the most common form.[6]

In humans, chromosome 21 (C21) is the smallest. Since humans have one pair of sex chromosomes (XX in females and XY in males) and 22 sets of autosomes, this chromosome should be labeled with the number 22.[7] This numerical error has not been rectified since the early days of molecular genetics when microscopes lacked the discriminatory capacity, they now possess. At the time, C21 was incorrectly placed in the chromosomal order of magnitude. The human C21 chromosome, sometimes

called Hsa21, has around 250 protein-coding genes and anywhere from 165 to 424 non-coding RNA. The genes that control gene expression (the actual number varies depending on genome annotation).[8]

While bicuspid aortic valve was the most frequent congenital abnormality in adults. Utmost infants born with a congenital heart problem have a ventricular septal defect (VSD). The fundamental cause of hemodynamic compromise in VSD is an irregular connection between the right and left ventricles and the creation of a shunt.[9] A ventricular septal defect (VSD) occurs when this structure doesn't close normally during embryonic and fetal development. The most common kind of congenital heart defect is a VSD, which may occur on its own or as part of a more complicated cardiac anomaly. This trait accounts for many cases of congenital cardiac disease.[10,11] Except for outlet VSDs, around 90% of minor VSDs close spontaneously within the first year of life. Closure occurs more often as children become older; rates start at 24% at 18 months, rise to 50% at 48 months, and peak at 75% at 120 months, according to research. If the VSD doesn't close on its own throughout infancy, it might remain present into adulthood without being noticed until a difficulty arises.[12]

METHODS

This descriptive research study was conducted cardiology unit of Shaikh Zayed Hospital, Lahore from March 2022 to August 2022. All 269 children with Down syndrome between the ages of 1 and 12 who were diagnosed and included, regardless of the presence or absence of symptoms, signs, x-ray chest, or ECG abnormalities that would point to CHD. Lesions with no substantial impact on hemodynamics, such as patent foramen ovale (PFO) and PDA less than 1 mm, were not included. All of these children had a complete blood count (CBC), serum electrolyte, renal function test, blood sugar, chest x-ray, regular urine examination, electrocardiogram (ECG), and other pertinent investigations performed after a thorough history and physical examination. Phenotypic findings such as mongoloid facies, brachycephaly, depressed nasal bridge, protruding tongue, small low set ears, upward slanted eyes with epicanthic fold, short neck, short and broad hands, transverse single palmar crease, clinodactyly, a large space between the toes (sandal gap), hypotonia, and delayed milestones were used to diagnose all children with DS.

Each item on the data collecting page was tallied, and the demographic information was calculated. IBM's Social Science Statistical Package, Version 20 (IBM SPSS, Armonk, NY, USA) was used to analyze the data. Frequency and percentage displays were used to depict the categorical data. According to the results of the Shapiro-Wilk test, the distribution of ages is not normally distributed, thus we utilized the median and the range to summarize the data. Chi-square test or Fisher's exact test (where anticipated count in any cell was less than 5) was used to compare two groups' proportions. When the p-value was less than 0, it was deemed to be significant.

RESULTS

Patients' ages varied widely, from 1 year to 12 years, on average (mean 3.12.7 years). Patients younger than two made up the vast majority (n=166, 61.7%). Table 1 shows that 211 (78.4%) patients were born at full term whereas 58 (21.6%) were delivered prematurely. The study population consisted of 143 (53.2% of the total) male and 126 (46.8% of the total) female patients.

Table 2 shows that 56 individuals with Down syndrome (20.8%) were diagnosed with a ventricular septal defect. As can be seen in Table 3, there was no statistically significant difference in the prevalence of VSD across the different subgroups defined by patient age (p=0.976), gender (p=0.711), or term/preterm status (p=0.735).

Table 1: Baseline Characteristics of Study Sample

Variables	Characteristics	Study Sample
Age (years)	Mean ± SD	3.1 ± 2.7
	≤2 years	166 (61.7%)
	2-5 years	67 (24.9%)
	>5 years	36 (13.4%)
Gender	Male	143 (53.2%)
	Female	126 (46.8%)
Delivery	Term	211 (78.4%)
	Preterm	58 (21.6%)

Table 2: Frequency of Ventricular Septal Defect in Children with Down Syndrome

Ventricular Septal Defect	Frequency (n)	Percent (%)
Yes	56	20.8
No	213	79.2
Total	269	100

Table 3: VSD Classification Based on a Number of Different Subgroups

	Characteristics	n	VSD n (%)	P value
Age	**≤2 years**	166	35 (21.1%)	0.976
	2-5 years	67	14 (20.9%)	
	>5 years	36	7 (19.4%)	
Gender	**Male**	143	31 (21.7%)	0.711
	Female	126	25 (19.8%)	
Delivery	**Term**	211	43 (20.4%)	0.735
	Preterm	58	13 (22.4%)	

DISCUSSION

The incidence of VSD in children with DS ranges between 40-63%, making it one of the primary causes of illness and death in this age group. Research suggests that the most often detected lesion in children with DS who have CHD varies throughout parts of the globe. Considering the high risk of pulmonary vascular disease in these patients, it is crucial to understand the profile and connection of CHD in DS for a certain geographic region in order to plan therapy such intervention, surgical care, and medical follow-up.[13] Down syndrome, also known as trisomy 21, was first identified in 1866. The remaining 5% are translocations (3% of all instances) and mosaics (2% of all cases). One in every 650 births is a presentation. About 1% of the population is affected. Cardiac abnormality is the leading cause of death in the first two years of life.[7,11]

This study's prevalence of CHD (21.6%) is in line with previous research. It's quite close to the 20.8% reported by Teteli *et al*.[14] in East Africa and the 22.0% reported by Martin *et al*. in the USA.[15] Congenital cardiac problems occur at a much greater rate than other birth defects. Researchers found a rate of 52.5% in Pakistan, but only 5.7% in Nigeria reported by Okeniyi *et al*.[21] and 12.7% in Mexico observed by Figueroa *et al*.[17] Possible causes for this variation include, but are not limited to, differences in embryological systems and national genetic makeup.

A total of 143 (53.2%) men and 126 (46.2%) women participated in the current research, for a male to female ratio of 1.1:1. Similarly, Ibrahim *et al*. (2012) in Sudan observed a male-to-female ratio of 1.1:1 among individuals with Down syndrome.[18] Kumar *et al*. (2017) found it to be 1.7:1 among Indian patients with Down syndrome, whereas Munsi *et al*. (2014) found it to be 1.5:1 in Bangladesh respectively.[20,19] While Okeniyi *et al*. (2017) found a female preponderance (m/f; 1:1.2) in Nigeria, Figueroa *et al*. (2003) found a 1:1 male-to-female ratio among Mexican patients with this condition.[20]

Twenty-eight percent of the people in this research who had Down syndrome also had a ventricular septal defect. Our finding agrees with the 22.6% frequency found by Khan *et al*. (2012) among patients coming to Lady Reading Hospital Peshawar.[7] The incidence of VSD in individuals with Down syndrome has been observed at comparable rates by groups in the USA observed by Martin *et al*., 22.0%[15] and in East Africa reported by Teteli *et al*., 20.8%.[14]

Consequently, VSD is a frequent birth abnormality that affects the heart in infants with Down syndrome. Doppler echocardiographic screening for this anomaly in children with Down syndrome should become standard practice in light of these findings since early detection and treatment may lead to better outcomes.

CONCLUSION

It is necessary to do more research in order to promote programs sufficient and cost-effective for the monitoring and care of diabetes patients, as poor glycemic control negatively impacts the efficacy of antiviral treatment in HCV RNA-positive patients.

LIMITATIONS

The data shown here is not representative of the whole population and comes from a single location. The diagnosis was made mostly on clinical grounds without the use of cytogenetic testing. Therefore, we were unable to make any statements on the prevalence of VSD in various down syndrome chromosomal changes.

SUGGESTIONS / RECOMMENDATIONS

Single center study doesn't accurately represent population-based incidence so it would be conducted in multiple centers.

CONFLICT OF INTEREST / DISCLOSURE

None.

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