

## Original Article

# Down's Syndrome with Congenital Heart Disease: Our Surgical Experience

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## Abstract

### Key words:

Down's syndrome, congenital heart disease, cardiac surgery.

**Background:** Our objective was to analyze the outcome of patients of Down's syndrome with congenital heart diseases undergoing cardiac surgery.

**Methods:** This was a retrospective study conducted between January 2013 and June 2019. 49 consecutive patients with Down's syndrome with congenital heart disease admitted in pediatric cardiac surgery unit at National Institute of Cardiovascular Diseases (NICVD). Patients were followed up postoperatively for in-hospital outcome.

**Results:** Among 49 patients the heart lesion ranked in incidence as follows- VSD 24(48.97%), AV canal defect 12(24.48%), TOF 6(12.24%), PDA 6(12.24%) and ASD 1(2.04%). Pulmonary hypertension was found in 63.25% patients. Moderate pulmonary hypertension was most common, found in 18(38.66%) patients. Severe and mild pulmonary hypertension was found in 10(32.38%) and 3(9.67%) patients respectively. All the patients had undergone surgical correction. The postoperative period was complicated in 44.89% of patients. The most frequent complication was pulmonary infection 20.40%, Wound infection 6.12% and low output syndrome 6.12% were the next. One patient had postoperative heart block, needed permanent pace maker implantation. In-hospital mortality was 12.24%.

**Conclusion:** Patients with Down's syndrome with congenital heart disease undergoing surgical correction had an acceptable postoperative morbidities and mortality.

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## Introduction:

Down's syndrome or trisomy 21, the most common human chromosome disorder predisposes affected individuals to a myriad of multisystemic manifestations and mental sub normality with an incidence between 1:700 and 1:800 live births.<sup>1,2</sup>

Congenital heart disease (CHD) is reported to occur in 40% to 60% of patients with Down's syndrome, with complete atrioventricular septal defect being the most common.<sup>3,4</sup> Other frequently occurring lesion includes ventricular septal defect (VSD), patent ductus arteriosus (PDA), atrial septal defect (ASD) and Tetralogy of Fallot (TOF) in decreasing order.<sup>5</sup>

Children with Down's syndrome who do not have CHD have much better out comes and early corrective surgery for those with CHD greatly improves life expectancy.<sup>6</sup>

Previous studies that evaluated outcomes after congenital heart surgery for patients with Down's syndrome showed conflicting results. Some reported increase mortality rates, length of hospital stay, and morbidities such as duration of ventilation and infection rates, where as other suggested similar or improved outcomes for patients with Down's syndrome who underwent atrio-ventricular septa defect repair.<sup>3,4,7,8</sup>

The purpose of these study was to describe perioperative outcome of patients with Down's syndrome with CHD undergoing cardiac surgery.

## Methods:

We conducted a retrospective study of Down's syndrome with congenital heart disease that

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underwent cardiac surgery from January 2013 to July 2019, at Pediatric Cardiac Surgery unit in NICVD. Data collection included demographic information, preoperative data, ECG, Echocardiogram, thyroid status and surgical procedures. Operative data included duration of cardiopulmonary bypass time, cross clamp time. Outcome data include hospital death, total postoperative length of hospital stay, postoperative complications such as infections, pulmonary complications, arrhythmia, atrioventricular block, pulmonary hypertensive crisis are noted.

Results: A total of 49 patients of Down's syndrome with CHD underwent surgery. Age of the patient at the time of surgery was 5 months to 13 years.

Male were 20 and female were 29 in numbers and weight of them was 4 kg to 29 kg. Patients characteristics were analyzed in Table I.

**Table-I**

*Distribution of patients by age (n=49).*

Age	Number of Patients
5 to 24 months	20
2 to 4 years	13
4 to 6 years	7
6 to 8 years	5
> 8 years	4

The 5 most common CHD found in patients with Down's syndrome are presented in Table II. VSD was the most common (48.97%) found in this study. Among them inlet type was most common.

**Table-II**

*Distribution of CHD in children with Down's syndrome (n=49).*

Type of CHD	Number (%)n=49
VSD	24 (48.97)
Inlet	12
Perimembranous	08
Doubly committed	04
AVSD	12 (24.48)
Partial	02
Transitional	03
Complete	07
PDA	06(12.24)
Large PDA	05
Moderate PDA	01
TOF	06(12.24)
ASD	01 (2.04)

43 of 49 patients presented with left to right shunt. Out of 43 patients of L-R shunt, 31 patients presented with pulmonary hypertension. Among which, moderate pulmonary hypertension was the most common. Table III showing the distribution of pulmonary hypertension in patients.

**Table-III**

*Distribution of pulmonary hypertension in patients (n=49).*

Type of CHD	Mild	Moderate	Severe
n= 31	PAH	PAH	PAH
VSD	02	11	04
PDA	00	03	02
AV Canal Defect	01	04	04
Total	3 (9.67%)	18 (58.06%)	10 (32.25)

14 (28.57%) patients presented with hypothyroidism.

Complete AV canal defect and TOF repair required longer CPB and cross clamp time. Table IV showing the distribution of operative characteristics.

**Table-IV**

*Distribution of operative characteristics (n=49).*

Procedure	CPB Time (minutes)	Cross Clamp Time (minutes)
ASD closure	44	21
VSD closure	58 (45-79)	24.83 (15-45)
AV canal defect repair	108 (60-165)	64 (35-100)
TOF repair	91 (80-160)	65 (55-95)

Regarding post-operative outcomes TOF repair patients required longer mechanical ventilation time and ICU stay than AV canal defect repair. VSD patients required comparatively short ventilation and hospital stay time. Table V showing the distribution of post-operative outcome.

Early postoperative pulmonary complications were more common. One patient required tube thoracostomy for pleural effusion. One TOF patient needed mediastinal re-exploration due to mediastinal hematoma. One patient of complete AV canal defect required permanent pace maker implantation. Other complications include arrhythmia in 2 patients, low output syndrome in 3 patients and renal failure in 1 patient. Table VI showing distribution of postoperative complications.

**Table-V**  
*Distribution of post-operative outcome (n=49).*

Type of surgery	Ventilation time (hour)	ICU stay (days)	Hospital stay (days)	Mortality
ASD closure	4	2	5	0
VSD closure	9 (6-24)	3 (2-4)	7 (5-10)	3
AV canal defect repair	14 (8-36)	3 (2-5)	8 (7-12)	1
TOF repair	24 (10-48)	5 (3-10)	9 (8-17)	2
PDA closure	2.5 (2-3)	1.5 (1-2)	3 (2-4)	0

**Table-VI**  
*Distribution of postoperative complications (n=49).*

Post-operative morbidity	Number (%)
Pulmonary infection	10 (20.40)
Wound infection	03 (6.12)
Pleural effusion	01 (2.04)
Low cardiac output syndrome	03 (6.12)
Renal failure	01 (2.04)
Arrhythmia	02 (4.08)
Heart block	01 (2.04)
Mediastinal haematoma	01 (2.04)

In-hospital mortality occurred in 6 (12.24%) patients. Among these 3 VSD patients were due to intractable pulmonary hypertensive crisis, septicemia and hyperpyrexia with convulsion.

### Discussion:

The earliest representation of Down's syndrome dates back to 1505. It was later described by Sequis 1846 and John Langdo Down in 1866.<sup>9</sup> The association of congenital heart disease and Down's syndrome was recognized by Garrod in 1894.<sup>10</sup> Patients with Down's syndrome tended to have reduced life expectancy in the context of multiple congenital anomalies involving the cardiovascular, pulmonary, gastrointestinal, hematological, endocrine, neurologic and immunologic systems. The advancement in medical and surgical care and de-institutionalization improved the survival of patients with Down's syndrome.<sup>11-14</sup> Congenital heart disease affecting almost half the patients with Down's syndrome. The purpose of our study to share experiences of surgical outcome of Down's syndrome with congenital heart disease who underwent for corrective cardiac surgery.

The types of heart defect with Down's syndrome may vary according to geographic region. In this

study the most common type was VSD (48.97%) which is similar to China population (40%)<sup>15</sup> but different from United States<sup>16</sup> and France<sup>17</sup> where atrioventricular septal defect was more frequent.

59.18% population of this study was female which is similar in Brazilian study 56.1%.<sup>18</sup> In evaluation of patient characteristics, we found that children with Down's syndrome were younger at the time of surgery for all procedures evaluated except the AV canal defect and TOF.

Pulmonary arterial hypertension (PAH) was recorded in 63.26% of the children with Down's syndrome. Shrestha reported 52.5% having PAH and Mourato recorded 37.5% developing PAH.<sup>19,20</sup> It has been reported that patient with Down's syndrome develop pulmonary hypertension early when they have left to right shunt lesions. Individuals with Down's syndrome may have pulmonary hypertension for various reasons such as chronic airway obstruction, abnormal growth of alveoli, and thinner pulmonary arterioli.<sup>21,22</sup>

In this study, nearly half of patients underwent VSD closure done by traditional manner. CPB and cross clamp time was as like of usually VSD closure without Down's syndrome but take a little longer time in complete A-V canal defect and TOF repair. Where all patients need transannular patch.

Patient with Down's syndrome undergone, TOF repair and A-V canal defect had significantly longer ventilation time and length of hospital stay compare with patient without Down's syndrome.<sup>23</sup>

Postoperative pulmonary infection was the most common complication affecting 20.40%, other reported 28%.<sup>24</sup> Malecand and his colleagues reported higher rates of post-operative complications including respiratory infections and sepsis, which led to prolonged ventilation and

longer length of ICU stay.<sup>25</sup> Chronic upper airway obstruction increased secretion and gastroesophageal reflux leading to chronic aspiration and concomitant immunodeficiency may play a role.<sup>26</sup>

A patient of complete AV canal defect repair had post-operative complete heart block requiring permanent pacemaker placement. Anomalies in the conductive system in patients with AV canal defect have been reported.<sup>27</sup>

In this study, there were 6 (12.24%) in-hospital deaths. Causes of death includes aspiration pneumonia, intractable pulmonary hypertensive crisis, high pyrexia with convulsion in VSD patients, arrhythmia in AV canal defect and low output syndrome with multi organ failure in TOF patients. FA Bacieuciz Jr and his colleagues reported mortality 16.40% and TOF was the highest.<sup>28</sup> There was trends towards increase mortality for patients with Down's syndrome undergoing TOF repair and decreased mortality rate for patients with Down's syndrome undergoing AV canal defect repair.<sup>23</sup>

### Conclusion:

Congenital heart disease is the most frequent association with Down's syndrome and remains a major cause of morbidity and mortality. Surgery plays a major role for better outcome with acceptable morbidity and mortality.

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### Conflict of Interest - None.

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