Survival Analysis of Down Syndrome with Congenital Heart Disease: A 5-years Registry at QSNICH

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Background: *Down syndrome (DS) is the most common chromosomal abnormality in children. Atrio-ventricular septal defect (AVSD) is the most common congenital heart disease (CHD) reported in DS. The mortality rate of DS with congenital heart disease (CHD) is 5-7 times higher than normal population. The survival rate in DS has improved with time and has reported up to 91% and 85% at one and ten years of age, respectively.*

Objective: *To study the prevalence of CHD, clinical course, treatment, the overall survival in patients with DS compare with those who are associated with CHD.*

Study Designs: *DS registry, multidisciplinary approach, single centre.*

Material and Method: *All DS patients at QSNICH with parental signed consent were enrolled in the registry. The study was conducted for 5 years starting from May 2007 to April 2012. All patients were followed-up according to schedule modified from American Academic of Pediatrics (AAP) health supervision guideline. Standard treatment was given to all those children with diagnosis of CHD and/or other associated diseases.*

Results: *Four hundred and two cases of DS were enrolled. Two cases were excluded due to the parental inconvenience. The mode and mean age of the patients at registration were 1 and 7 months (1-62). Two hundred and seventy-one cases had an initial echocardiographic diagnosis of CHD, which included 91 of Patent ductus arteriosus (PDA), 49 of Ventricular septal defect (VSD), 34 of AVSD, 34 of secondum Atrial septal defect (ASD), 6 of Tetralogy of Fallot (TOF), 2 of Coractation of Aorta (CoA), 11 of other CHD and 44 of combined lesions. During the follow-up period, spontaneous closure of PDA, VSD and ASD occurred in 46, 12 and 15 cases, respectively. After the exclusion of those who had spontaneous closure, the prevalence rate of CHD in DS was 49.8%. VSD was the most common lesion, slightly more than PDA. Ninety-eight cases underwent cardiac surgery; including 39 of associated VSD, 24 of AVSD, 26 of PDA, 3 of TOF, 4 of CoA/AA repaired, one each of Cor triatriatum and primum ASD. Twelve cases had catheter interventions, including PDA occlusion (10 cases), and 1 case of balloon dilatation of re-CoA and coils embolization of isolated major aorto-pulmonary collateral artery. Seven cases were lost at follow-up. By using the hospital information and National Health Security Office (NHSO) database, 47 infants died during the follow-up period. The causes of death were immediate post cardiac surgery in 10 cases (AVSD in 9, VSD in 1) and not associated with surgery in 37 cases including CHF in 8, pneumonia in 7, leukemia in 4, airway disease in 3, others 7 and unknown 8 cases. The overall survival at 1and 5 years of age were 96% and 86%, respectively. DS with CHD had a significantly lower survival rate than those without CHD ($p<0.001$).*

Conclusion: *According to this study, the prevalence rate of CHD in DS was high. VSD was the most common lesion and has better prognosis than AVSD. The main cause of death was a cardiac problem. Follow-up patients by using modified AAP guideline and standard treatment can improve their overall survival.*

Keywords: *Down syndrome, Congenital heart disease, Survival, Risk factor*

J Med Assoc Thai 2014; 97 (Suppl. 6): S108-S114
Full text. e-Journal: *http://www.jmatonline.com*

Down syndrome (DS) is the most common chromosomal abnormality in children. There are multiple major congenital anomalies in DS[1,2]. Heart disease is the most common congenital abnormality and various types of congenital heart diseases (CHD) have been reported in DS[3]. Atrio-ventricular septal defect (AVSD) was the most common CHD in DS[4-6], followed by ventricular septal defect (VSD) and others. Life expectancy in DS is shorter than normal population, especially if it is associated with significant CHD[7]. The mortality rate in DS is 5-7 times higher than

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normal[8,9] when associated with CHD. The survival rate of DS is improving with time[7,10-12]. This could be due to the multidisciplinary approach and specific congenital anomalies treatment especially surgical correction of congenital heart disease[13]. One report has shown the survival rate in DS to be 91% at one year and 89% at ten years of age[14]. However, there have been no long-term survival rates reported from Thailand. The objectives of the present study were to find out the prevalence rate of CHD and the survival rate of DS patients with and without CHD in Thailand.

Material and Method

The design study was a DS registry with multi-disciplinary approach. A programmed follow-up of the patients was modified from the AAP guideline[15], which is shown in Table 1. The enrollment period was from May 2007 to April 2012 at Queen Sirikit National Institute of Child Health (QSNICH). All patients with DS attending any clinic at QSNICH during the study period were included in the present study after obtaining informed consent from the parents. Complete cardiac work-ups including echocardiogram were performed in all cases at the first cardiology visit, if possible. Standard treatments were used for those with abnormal diagnostic findings. The cardiology follow-up schedules were every 1-2 months if the patients were symptomatic and every 6 months if they were asymptomatic or were not scheduled for any cardiac interventions during that period. Phone calls were made and the patients to ascertain information in case they did not return for their scheduled follow-up. Hospital information systems were also searched for the last visit to the other departments if they could not be reached by phone calls. Survival of some patients who did not return for follow-up was checked from the network of national health security office (NHSO) of Thailand. Kaplan-Myer analysis was used for survival rates and curves.

Results

During the 5-year study period, 402 cases of DS were enrolled. Two cases were excluded due to the parental inconvenience; 400 cases of DS were followed-up. Ages at the first enrollment ranged from less than 1 month to 62 months. The mean age at presentation was 7 months with mode at 1 month as shown in Fig. 1. The results of the first echo diagnosis showed that 271 patients had CHD (Table 2). Forty-six cases of patent ductus arteriosus (PDA), 12 cases of ventricular septal defect (VSD) and 15 cases of atrial septal defect (ASD) had spontaneously closed. One hundred and ninety-nine cases had persistent cardiac defects, giving the prevalence rate of CHD in this case series equal to 49.8%. One hundred and sixty cases had isolated lesions and 39 cases had multiple cardiac lesions. After the spontaneous closure, VSD and its associated lesions was the most common diagnosis slightly more than PDA (73 vs. 72 lesions) and about 1.9 times more than AVSD. All AVSD cases were the complete type. Twenty-four cases of VSD were classified as a perimembranous inlet VSD and there were two cases of isolated primum ASD, which have been classified by many as partial AVSD.

Table 1. Modified AAP guideline for health supervision for children with Down syndrome

	Genetic									
	Ch.study CBC	PE, HC growth	TFT	Cardio	Eye	Ear	Resp	Dent	Ortho	Dev
Newborn/ 1st visit	Y	Y	Y	Y	Y	Y				Y
6 m		Y	Y		Y		Y			Y
12 m		Y	Y		Y		Y	Y		Y
24 m		Y	Y		Y		Y	Y		Y
36 m		Y	Y		Y		Y	Y	Y	Y
48 m		Y	Y		Y		Y	Y	Y	Y
60 m		Y	Y		Y		Y	Y	Y	Y

Ch.study = chromosome study; CBC = complete blood count; PE = physical examination; HC = head circumference; TFT = thyroid function test; Resp = respiratory system; Dent = dentist; Ortho = orthopedics; Dev = development; Y = should be performed

During the follow-up period, 98 cases (49.2%) had cardiac surgery and 12 cases underwent catheter intervention. All, except three cases had corrective procedures. The indications for surgery were CHF, severe cyanosis not responding to medication or poor weight gain. Small to moderate size PDA had elective trans-catheter closure with coils or devices after one year of age. At the last follow-up, seven cases were still on medication, 3/7 on low dose aspirin for modified Blalock-Taussig shunt (BTS). Most cases were categorized as functional class I, except for four cases who refused surgery or was inoperable due to late presentation, who were in functional classes II-III. Seven cases were lost in follow-up.

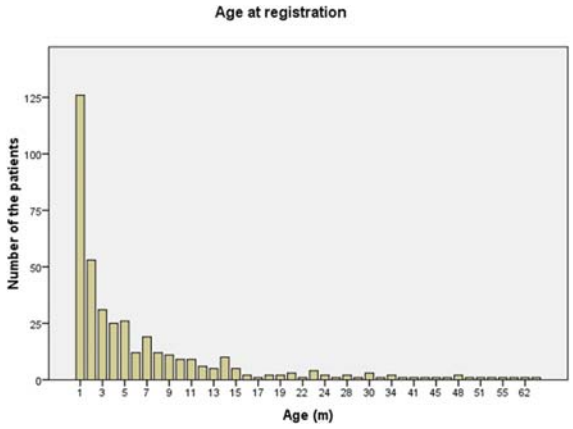

Fig. 1 Age distribution at registration and number of the patients.

The overall survival at 1 and 5 years of age were 96% and 86%, respectively. The Kaplan-Meier survival curve (Fig. 2) showed that the survival rate slowly dropped with age but remained steady at about 40 months of age. Comparison of the survival rate of DS with and without CHD (Fig. 3) showed that there was a difference in survival rate of the patient DS with and without CHD before the first year of life, which stabilized after two years of age. At 1 and 5 years of age, the survival rate of DS without CHD was 99% and 92% as compared to 91.3% and 79.6% of those with CHD, respectively.

Forty-seven patients died during the follow-up period. Seven point nine percent (16/201) of DS without CHD and 15.6 percent (31/199) of DS with CHD died. CHD was a major risk factor of death in DS with the Hazard risk of 3.50; 95%CI 1.78-6.89. The causes of death were immediate post cardiac surgery, 10 cases (AVSD 8, VSD 1, DKS/BDG 1), congestive heart failure

Table 2. Prevalence of congenital heart disease diagnosis at initial echocardiogram, after spontaneous closure and the number of surgical and catheter intervention

Lesions	Initial echo (n)	SC (n)	After SC (n)	Surgery (n)	Catheter (n)
VSD	49	6	43	17	
VSD, ASD	8		8	6	
VSD, PDA	20	5	15	11	
VSD, PDA, ASD	4		4	3	
VSD, PDA, COA	1		1	1	1
VSD hypo AA	1		1	1	
VSD, COCM	1	1	1		
PDA	91	45	46	24	10
PDA, ASD	4	1	3	2	
AVSD	34		34	21	
AVSD, PDA	3		3	3	
ASD	34	15	19		
TOF	6		6	3	
CoA	2		2	2	
Prim. ASD	2		2	1	
AVSD, ToF	2		2	1	
Others	9		9	2	1
Total (cases)	271	73	199	98	12

VSD = ventricular septal defect; ASD = atrial septal defect; PDA = patent ductus arteriosus; CoA = coarctation of the aorta; hypo AA = hypoplastic aortic arch; COCM = congestive cardiomyopathy; AVSD = atrio-ventricular septal defect; TOF = tetralogy of Fallot; Prim ASD = primum atrial septal defect; SC = spontaneously closure

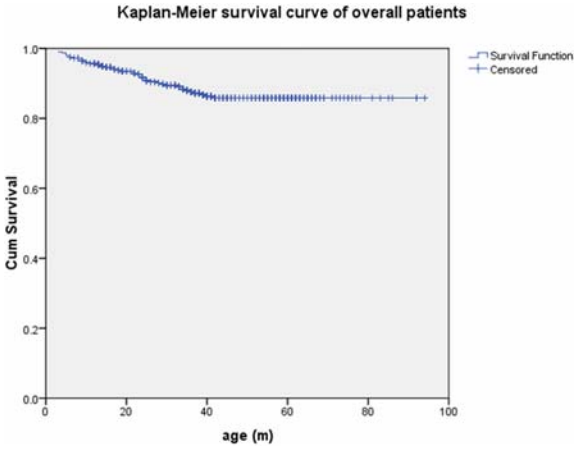

Fig. 2 Kaplan-Meier survival curve of overall Down syndrome patients.

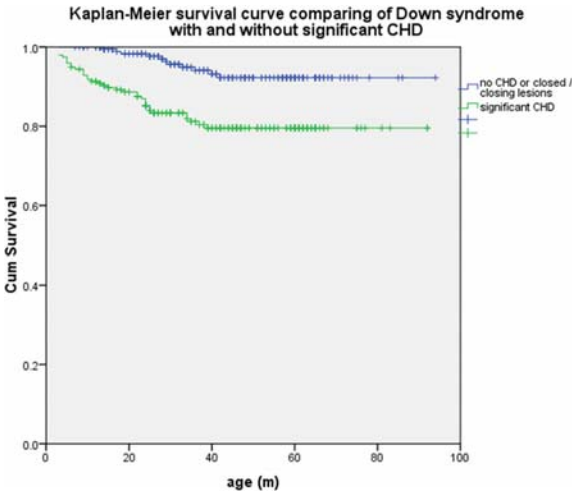

Fig. 3 Kaplan-Meier survival curve comparison between Down syndrome with and without significant congenital heart disease.

8 cases, pneumonia 7 cases, leukemia 4 cases, airway problems 3 cases, the other causes 7 cases and unknown 8 cases.

Discussion

The present report is a large series of DS patients who were registered at QSNICH since 2007 and had a medium term follow-up. Most cases presented in early infancy period, however, 15% of the cases registered after 1 year of age. Echocardiogram at the time of registration showed the prevalence rate of CHD was 67.7%. PDA was the most common diagnosis. After exclusion, those who had spontaneous closure during follow-up period, the prevalence of CHD in DS decreased to 49.8%. This prospective echocardiogram study showed a higher prevalence of CHD than the previous report from Thailand[3]. Isolated VSD occurred frequently less than isolated PDA but when each was counted as part of multiple lesions, VSD was slightly more prevalent than PDA and 1.9 times more frequent than AVSD. However, AVSD may be considered more prevalent than VSD (63 vs. 49 cases), if the perimembranous-inlet type of VSD and primum ASD are considered a partial form of AVSD. Since QSNICH is a tertiary care hospital, most of the DS cases had been referred from all over Thailand for investigation and treatment. The prevalence rate of CHD in the present study may be higher than general DS population.

The present study showed that the overall survival rate of DS was 96% and 86% at 1 and 5 years, respectively. This survival rate was slightly different from the developed countries. Compared to other previous studies[14,16], the survival rate at 1 year in this study was higher (96% vs. 91-92.9%). This may be due to the standard protocol of early surgery within the first year of life, which may cause a higher 1-year mortality rate but better long-term survival. The long term survival rate at 5 year (86%) in this present study is lower than the survival rate at 10 year of 85-88.6% from the previous studies[14,16]. Patients with major CHD need either surgery or cardiac interventions very soon after birth with complete repair within the 1st year of life to avoid late complications of recurrent lung infection, severe pulmonary hypertension or pulmonary vascular obstructive disease, which may be the cause of poor outcomes if surgery is performed later on. From the survival curve in Fig. 2, the survival rate falls until about 3.5 years of age.

CHD is the risk factor of survival in DS patients[7,11,16,17] and also the risk factor for lower respiratory tract infection[18]. The survival rate was higher in a group of DS without CHD when compare to those of DS with CHD. From Kaplan-Meier survival curve, comparison of DS with and without CHD (Fig. 3) showed the higher survival in DS without CHD. The difference in survival of both groups started within the first year of life and increased to about three years of age after which it stabilized. Thus, in the present study CHD is a risk factor in DS on the mortality during infancy. The difference of 12-13% in survival rates between these two groups was significant, (p<0.001). This present demonstrated that CHD in DS increases the risk of mortality rate by 3.5 compare to those without CHD.

The mortality rate was high either from pre or

post cardiac operation, especially in patients with complete AVSD. Fifteen patients died from congestive heart failure and pneumonia prior to the operation. Many of these cases had been referred with complicated lungs infection before the diagnosis of serious CHD was made. Some needed prolonged respiratory support and treatment until resolution of lung infection before further intervention or surgery. The long waiting list for cardiac surgery is another determining factor in a developing country like Thailand, which has limited resources. Patients with better prognosis, simple lesions, less associated anomalies will be the first priority for surgery. Ten cases died immediately post operation; eight had AVSD. Twenty-five of 39 AVSD cases underwent cardiac repair during the study, showing 32 percent mortality rate in the repaired AVSD patients. This high rate is a cause for concern and indicates that there is need for improvement is the care of these patients.

Before and immediately after cardiac surgery in this study, loss of follow-up was due to inactive cardiac disease, lack of transportation, financial issues and poor parental attitude. The present study was designed to maximize the survival data by many methods. One is asking for more than two phone numbers from either the parents or their relatives at the enrollment. Another is to search the hospital information system for the last visit or next appointment to any of the departments in the hospital if failed phone calls. Lastly, if they did not contact to the hospital, the survival case was then searched from the National Health Security Office database via Thai ID number. Age of the surviving patient at the last contact or age at death was used for Kaplan-Myer survival analysis and curve. During the 5-year follow-up period, we were unable to gather information for seven patients.

There was one interesting case of DS with diagnosis of left to right shunt at small ASD with severe pulmonary hypertension. Clinically, he was thin, had poor weight gain and developed recurrent lung infection requiring frequent hospitalization. Several echocardiograms failed to explain his clinical problems. Serum immunoglobulin levels were normal and anti-HIV non-reactive. Cardiac CTA at 16 months of age showed an isolated big MAPCA, single supply to left lower lung. After multiple coils embolization of this MAPCA, there was much improvement to his clinical status.

A significant CHD in DS may have less clinical signs of CHD compared to non-DS with CHD. This may be due to frequent association with recurrent respiratory tract infection[19], early development of pulmonary vascular obstructive disease or secondary pulmonary hypertension from respiratory problems[20] in DS patient, which may have an effect on physical findings and timing of surgery. Some late presentation cases may go beyond surgery[21]. The golden period for surgical intervention differs with each type of CHD. Many cases were referred for treatment of other organ anomalies and found to have unexpected severe CHD. The modified health supervision from the AAP guideline[15] is helpful for a systematic multidisciplinary approach. All organ systems will be prospectively evaluated and receive treatment at the appropriate time. This may not only improve the survival rate, but also the quality of life in DS patients.

Potential conflicts of interest

None.

References

1. Abbag FI. Congenital heart diseases and other major anomalies in patients with Down syndrome. Saudi Med J 2006; 27: 219-22.
2. Kava MP, Tullu MS, Muranjan MN, Girisha KM. Down syndrome: clinical profile from India. Arch Med Res 2004; 35: 31-5.
3. Jaruratanasirikul S, Soponthammarak S, Chanvitan P, Limprasert P, Sriplung H, Leelasamran W, et al. Clinical abnormalities, intervention program, and school attendance of Down syndrome children in southern Thailand. J Med Assoc Thai 2004; 87: 1199-204.
4. Park SC, Mathews RA, Zuberbuhler JR, Rowe RD, Neches WH, Lenox CC. Down syndrome with congenital heart malformation. Am J Dis Child 1977; 131: 29-33.
5. Bhatia S, Verma IC, Shrivastava S. Congenital heart disease in Down syndrome: an echocardiographic study. Indian Pediatr 1992; 29: 1113-6.
6. Wells GL, Barker SE, Finley SC, Colvin EV, Finley WH. Congenital heart disease in infants with Down's syndrome. South Med J 1994; 87: 724-7.
7. Mastroiacovo P, Bertollini R, Corchia C. Survival of children with Down syndrome in Italy. Am J Med Genet 1992; 42: 208-12.
8. Nahar R, Kotecha U, Puri RD, Pandey RM, Verma IC. Survival analysis of Down syndrome cohort in a tertiary health care center in India. Indian J Pediatr 2013; 80: 118-23.
9. Miller A, Siffel C, Lu C, Riehle-Colarusso T, Frias JL, Correa A. Long-term survival of infants with atrioventricular septal defects. J Pediatr 2010; 156:

994-1000.
10. Mikkelsen M, Poulsen H, Nielsen KG. Incidence, survival, and mortality in Down syndrome in Denmark. Am J Med Genet Suppl 1990; 7: 75-8.
11. Hijii T, Fukushige J, Igarashi H, Takahashi N, Ueda K. Life expectancy and social adaptation in individuals with Down syndrome with and without surgery for congenital heart disease. Clin Pediatr (Phila) 1997; 36: 327-32.
12. Yang Q, Rasmussen SA, Friedman JM. Mortality associated with Down's syndrome in the USA from 1983 to 1997: a population-based study. Lancet 2002; 359: 1019-25.
13. Parvathy U, Balakrishnan KR, Ranjith MS, Saldanha R, Sai S, Vakamudi M. Surgical experience with congenital heart disease in Down's syndrome. Indian Heart J 2000; 52: 438-41.
14. Leonard S, Bower C, Petterson B, Leonard H. Survival of infants born with Down's syndrome: 1980-96. Paediatr Perinat Epidemiol 2000; 14: 163-71.
15. American Academy of Pediatrics: Health supervision for children with Down syndrome. Pediatrics 2001; 107: 442-9.
16. Rasmussen SA, Wong LY, Correa A, Gambrell D, Friedman JM. Survival in infants with Down syndrome, Metropolitan Atlanta, 1979-1998. J Pediatr 2006; 148: 806-12.
17. Shin M, Kucik JE, Correa A. Causes of death and case fatality rates among infants with down syndrome in metropolitan Atlanta. Birth Defects Res A Clin Mol Teratol 2007; 79: 775-80.
18. Bloemers BL, van Furth AM, Weijerman ME, Gemke RJ, Broers CJ, van den EK, et al. Down syndrome: a novel risk factor for respiratory syncytial virus bronchiolitis—a prospective birth-cohort study. Pediatrics 2007; 120: e1076-e1081.
19. Hilton JM, Fitzgerald DA, Cooper DM. Respiratory morbidity of hospitalized children with Trisomy 21. J Paediatr Child Health 1999; 35: 383-6.
20. Miguel-Diez J, Villa-Asensi JR, Alvarez-Sala JL. Prevalence of sleep-disordered breathing in children with Down syndrome: polygraphic findings in 108 children. Sleep 2003; 26: 1006-9.
21. D'Alto M, Mahadevan VS. Pulmonary arterial hypertension associated with congenital heart disease. Eur Respir Rev 2012; 21: 328-37.

อัตราการอยู่รอดของเด็กกลุ่มอาการดาวน์ที่มีโรคหัวใจพิการแต่กำเนิด: ผลการศึกษา 5 ปี จากการลงทะเบียนผู้ป่วยที่สถาบันสุขภาพเด็กแห่งชาติมหาราชินี

ธนะรัตน์ ลยางกูร, ธวัชชัย กิระวิทยา, ชัยสิทธิ์ แสงทวีสิน, วรการ พรหมพันธุ์, พิมพ์ภัค ประชาศิลป์ชัย, อมรรัตน์ เพชรดำรงค์สกุล

ภูมิหลัง: กลุ่มอาการดาวน์เป็นโรคความผิดปกติของโครโมโซมที่พบได้บ่อยที่สุด ความพิการแต่กำเนิดของหัวใจที่รายงานว่าพบบ่อยสุดคือ Atrio-ventricular septal defect (AVSD) โรคหัวใจในเด็กกลุ่มอาการดาวน์เป็นสาเหตุที่ทำให้เด็กเสียชีวิตเพิ่มขึ้น 5-7 เท่า อัตราการอยู่รอดของเด็กกลุ่มนี้ดีขึ้นเรื่อยๆ จากการศึกษาที่ผ่านมาในประเทศที่เจริญแล้วพบอัตราอยู่รอดที่ 1 ปี และที่ 10 ปีสูงถึงร้อยละ 91 และ 85 ตามลำดับ

วัตถุประสงค์: เพื่อศึกษาอุบัติการณ์ของโรคหัวใจ การดำเนินโรค ผลการรักษา และอัตราการอยู่รอดของเด็กกลุ่มอาการดาวน์ที่มีและไม่มีโรคหัวใจ

วัสดุและวิธีการ: การลงทะเบียนผู้ป่วยเด็กกลุ่มอาการดาวน์ และการรักษาแบบสหสาขาวิชาชีพ เด็กกลุ่มอาการดาวน์ ทุกรายที่เริ่มมารับการรักษาครั้งแรกที่สถาบันสุขภาพเด็กแห่งชาติตั้งแต่ เดือนพฤษภาคม พ.ศ. 2550 ถึง เดือนเมษายน พ.ศ. 2555 ทุกรายจะได้รับคำแนะนำและเซนต์ใบยินยอมเข้ารับการลงทะเบียนให้การตรวจค้นหาความผิดปกติ แบบสหสาขาวิชาชีพและติดตามโดยการปรับรูปแบบ จากคำแนะนำของราชวิทยาลัยกุมารแพทย์ของประเทศสหรัฐอเมริกา เมื่อตรวจพบความผิดปกติไม่ว่าทางด้านโรคหัวใจหรือด้านอื่นร่วมด้วยเด็กทุกรายจะได้รับการรักษาแบบมาตรฐาน

ผลการศึกษา: เด็กที่เข้าร่วมโครงการเมื่อเริ่มต้นจำนวน 402 ราย แต่มีอยู่ 2 รายที่บิดามารดาไม่พร้อมที่จะเข้าร่วมโครงการ จึงเหลือเด็กกลุ่มอาการดาวน์ทั้งสิ้น 400 ราย ได้รับการติดตามดูแลรักษา อายุที่เริ่มต้นเข้ารับการลงทะเบียนโดยเฉลี่ยอายุที่ 7 เดือน (1-62 เดือน) เด็กจำนวน 271 ราย ที่ตรวจจากคลื่นเสียงสะท้อนหัวใจเมื่อเริ่มต้นว่ามีความผิดปกติทางด้านหัวใจ ได้แก่ PDA 91 ราย VSD 49 ราย AVSD 34 ราย secondum ASD 34 ราย TOF 6 ราย CoA 2 ราย โรคหัวใจอื่นๆ 11 ราย และโรคหัวใจที่มีหลายอย่างร่วมกัน 44 ราย ตลอดระยะเวลาที่ติดตาม 5 ปี พบว่ารูรั่วหลายชนิดที่ปิดได้เองตามธรรมชาติ ได้แก่ PDA, VSD และ ASD จำนวน 46, 12 และ 15 ราย ตามลำดับ หลังจากที่หักกลุ่มนี้ออกไปแล้วพบว่า มีอุบัติการณ์ของโรคหัวใจพิการแต่กำเนิด 49.8% (199 ราย) พบว่า VSD เป็นโรคที่พบบ่อยที่สุดมากกว่า PDA เล็กน้อย ได้รับการผ่าตัดโรคหัวใจจำนวน 98 รายได้แก่ VSD 39 ราย AVSD 24 ราย PDA 26 ราย modified BT shunt 3 ราย CoA, AA repaired 4 ราย Cor triatriatum และ primum ASD อย่างละ 1 ราย ได้รับการสวนหัวใจเพื่อการรักษา 12 รายได้แก่ปิด PDA 10 ราย ใช้ลูกโป่งขยาย re-CoA 1 ราย และอุด MAPCA ด้วยขดลวด 1 ราย จากการใช้ข้อมูลของสถาบันฯ ข้อมูลของสำนักงานหลักประกันสุขภาพแห่งชาติ มีเด็กที่ไม่สามารถติดตามได้ 7 ราย มีเด็กที่เสียชีวิตทั้งสิ้น 47 รายสาเหตุจากหลังการผ่าตัดโรคหัวใจ ขณะอยู่ในโรงพยาบาล 10 ราย (AVSD 8, VSD 1, DKS/BDG 1) ภาวะหัวใจวาย 8 ราย ปอดบวม 7 ราย มะเร็งเม็ดเลือดขาว 4 ราย โรคของหลอดลม 3 ราย โรคอื่น 7 ราย และไม่ทราบสาเหตุ 8 ราย อัตราการอยู่รอดของเด็กทั้งหมดที่ 1 ปี และที่ 5 ปีเท่ากับร้อยละ 96 และ 86 ตามลำดับเด็กที่มีโรคหัวใจมีอัตราการอยู่รอดต่ำกว่าเด็กที่ไม่มีโรคหัวใจอย่างมีนัยสำคัญ ($p<0.001$)

สรุป: จากการศึกษาพบว่าอุบัติการณ์ของเด็กโรคหัวใจในเด็กกลุ่มอาการดาวน์สูง 49.8% พบ VSD มากที่สุดใกล้เคียงกับ PDA และสูงกว่า AVSD 1.9 เท่าพบว่า VSD มีพยากรณ์โรคที่ดีกว่า AVSD สาเหตุของการเสียชีวิต ส่วนใหญ่เกิดจากโรคหัวใจ การดูแลรักษาติดตามโดยใช้การปรับเปลี่ยนแนวทางจากคำแนะนำของ ราชวิทยาลัยกุมารแพทย์สหรัฐอเมริกาและการรักษาแบบมาตรฐานจะช่วยให้อัตราการอยู่รอดดีขึ้น