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The Incidence of cardiac lesions in infants born with major gastrointestinal malformations in Northern Ireland

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SUMMARY

There is a recognised association between major gastrointestinal (GI) malformations and congenital heart disease (CHD). A retrospective study over 10 years involving 240 infants born with gastrointestinal malformations was conducted in the Royal Belfast Hospital For Sick Children (RBHSC). We felt it was important to look at the incidence of CHD diagnosed in the infants presenting to the tertiary referral centre in Belfast. Comparable figures for the incidence of CHD associated with major GI malformations was found in the literature.

INTRODUCTION

The association between major GI malformations and CHD is well recognised. The Royal Belfast Hospital For Sick Children is the regional referral centre for surgery in all infants born in Northern Ireland with major GI malformations. There are about 26,000 live births in Northern Ireland per year. Three hundred new cases of CHD are seen by the paediatric cardiologists, and about thirty infants are born with major GI malformations in the same period. Over the past 10 years all babies presenting with major GI problems have been referred for a cardiological assessment. This paper reports the outcome of that survey.

PATIENTS AND METHODS

All infants referred to the RBHSC with major GI malformations (diaphragmatic hernia, tracheo-oesophageal atresia/oesophageal atresia, anorectal anomalies, duodenal atresia, exomphalos and gastroschisis) between January 1989 and December 1998 were included in the study. All were seen by a paediatric cardiologist, usually prior to GI operation and each patient had an electrocardiogram (ECG), chest X-ray (CXR) and echocardiogram carried out. Since the majority of patients were assessed early in life, many had minor cardiovascular abnormalities, such as haemodynamically insignificant ductus arteriosus, patent foramen ovale and trivial pulmonary artery stenosis. These infants were not included as having a major cardiovascular abnormality.

RESULTS

In total, 240 infants with gastrointestinal malformations were identified over the 10 year period. There were 145 males and 95 females. Average gestation was just over 37 weeks and the mean birth weight 2480 grams. Of these, 37 (15%) had a recognisable syndrome such as Downs and Vater syndromes. Of the total of 240 infants, 53 (22%) had a congenital heart disease (table). Out of the 37 infants with recognised syndromes, a total of 24 (65%) had significant cardiac lesions and of the remaining 203 without syndromes, 29 (14%) had CHD.

The commonest GI abnormality among the infants without recognisable syndromes was imperforate anus (53 cases) and 12 (23%) of these were found to have congenital heart disease. The second commonest abnormality in this group was oesophageal atresia combined with tracheo-oesophageal fistula (32 cases); none had a recognisable syndrome and four (12%) had congenital heart disease.

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TABLE

Distribution of CHD with GI malformations

GI malformations	*No of Infants*	*No(%) with CHD*	*VSD*	*ASD*	*PDA*	*AVSD*	*FT*	*CoAo*	*TGA*	*PA*	*DORV*	*TA*	*Others*
Dia Hernia	35	6(17)		1	3				2				
TOF/OA	48	13(27)	3		3		1	1		1	1	2	Univentricular heart
Anorectal anomaly													
High	12	2(17)	2										
Low	57	14(25)	5	3	1				1	2	1		Hypoplastic left heart
Duodenal atresia	25	13(52)	4	1	1	6							Ebsteins anomaly
Exomphalos	22	4 (19)	2	2									
Gastroschisis	40	1(2)			1								
Total	240	53(22)	16	7	9	6	1	1	3	3	2	2	3

VSD = ventricular septal defect; ASD = atrial septal defect; PDA = Patent ductus arteriosus; AVSD = atrioventricular septal defect; FT = tetralogy of Fallot; Co Ao = coarctation of the aorta; TGA = transposition of the great arteries; PA = pulmonary atresia; DORV = double outlet right ventricle; TA = truncus arteriosus; TOF/OA = tracheo-oesophageal fistula/oesophageal atresia.

Of the total number of infants identified, only one had a family history of GI malformation (anal atresia in a sibling of an infant born with imperforate anus) and four with a positive family history of congenital heart disease, although only two of these were diagnosed as having significant cardiovascular abnormalities.

Fifty-five (23%) of the GI abnormalities were detected on antenatal scanning, the majority being large anterior abdominal wall defects, while only one infant with Downs syndrome and duodenal atresia had an atrioventricular septal (A-V) defect identified in utero.

223 (93%) were seen within the first week of life. Of the remaining 17 children seen after the first week, two had major cardiovascular abnormalities discovered at 10 and 19 days respectively.

Among those infants with known syndromes there was a total of 12 with VATER syndrome, (an acronym for *V*ertebral defects, *A*nal atresia, *T*racheo-*E*sophageal fistula, *R*enal defects and *R*adial anomalies), 15 with Downs syndrome and 10 others. All of the infants diagnosed with VATER syndrome had associated oesophageal atresia and tracheo-oesophageal fistula and five had coexisting imperforate anus. Of the 15 infants with Downs syndrome, the commonest GI abnormality was duodenal atresia in 11 infants, 10 of whom had associated congenital heart disease.

Of those transferred for corrective GI surgery, 42 (17%) of the 240 infants died, eight (19%) of whom had associated syndromes. Of the remaining deaths, five could be attributed to their CHD, while 29 (including eighteen with diaphragmatic hernia) were attributable to major uncorrectable GI malformations.

DISCUSSION

It has long been recognised that there is an association between major GI malformations and CHD; about 20% of patients with major GI malformations have an associated congenital heart defect.[1] In the 1970's, Greenwood *et al* reported the incidence of CHD in infants with a diagnosis of congenital diaphragmatic hernia, imperforate anus, exomphalocele and tracheo-oesophageal

fistula associated with oesophageal atresia to be 23%, 12%, 19.5% and 15% respectively.[2-5] Figures in this paper are 17%, 23%, 19% and 12%.

The recognition of a cardiac lesion in this group of patients is important, particularly in those with associated syndromes. Before embarking on major GI surgery it is important to be aware of the infant's cardiac status both for prognostic purposes and to provide sub-acute bacterial endocarditis (SBE) prophylaxis if appropriate. In a child with coexisting major GI and cardiac abnormality, it may be unwise to proceed with GI surgery. There is also a need to counsel parents regarding risks of future pregnancies. It has long been advocated that echocardiograms should be carried out on these infants as a matter of routine and that clinical examination, CXR and ECG alone, are not adequate to detect all the infants with CHD.[1]

Early reporting by parents of a family history of either CHD or major GI malformations is often sketchy although with an incidence of CHD in the new-born population at about 0.4-0.8%, our results seem to be representative.[7, 8]

Previous studies have revealed a much greater yield in the antenatal detection of large GI malformations compared with cardiovascular abnormalities.[9] Obviously with improvements in fetal ultrasonography, more GI malformations are being detected in utero; this compares to only one cardiovascular abnormality detected on scan in an infant with other congenital abnormalities. This is not surprising, since there is generally less difficulty in identifying large GI abnormalities compared to structural heart defects on antenatal scan. The number of GI malformations detected antenatally in this series appears to be low (only 23%). This may be slightly misleading since this information was taken from the paediatric rather than the antenatal notes. It is an important issue however, since mothers awaiting the birth of a child with a major GI malformation should be transferred to a regional centre capable of carrying out the necessary neonatal surgery.

The importance of early cardiac assessment is seen in the fact that two infants were operated on without echocardiogram and were not thought to have significant CHD (secundum atrial septal defect and a moderate-sized patent ductus arteriosus).

Although the overall incidence of CHD in association with GI malformations in our series of 240 infants was similar to other studies at 22%, the incidence of CHD amongst infants with recognisable syndromes and GI malformations was 65%. Careful examination is essential to rule out the possibility of an associated syndrome in an infant born with a GI malformation and these infants should be scanned early to rule out the high incidence of an associated cardiac defect. A previous study carried out in Belfast showed that infants with Downs syndrome had a 42% chance of having an associated cardiac abnormality, but in our series this is increased to 73% if there is a coexisting GI malformation.[10] This may suggest a greater risk of CHD in Downs syndrome if a GI abnormality is also present. VATER and associated conditions have a high risk of CHD occurring in infants with GI malformations.[1, 11, 12] Again, the infants with VATER association in our series with an incidence of 67% would concur with those previously reported.[13] Only one of the four infants with Beckwith-Wiedemann syndrome was found to have an associated cardiovascular abnormality. Previous studies have shown an association with isolated reversible cardiomegaly but not CHD.[14-17]

Most of the major GI malformations are correctable. The exception are infants with congenital diaphragmatic hernia, known to be associated with high mortality rates.[2, 18-21] Recent overall survival rates by the Congenital Diaphragmatic Hernia Study Group (formed in 1995 with sixty-two centres and 442 patients) was 62%.[22] Of the 35 infants identified with congenital diaphragmatic hernia over 10 years in our study, 17 (49%) survived. The figures for mortality in this paper are, however, not wholly representative, since severe cases with major bowel and cardiac problems never make it as far as the Children's Hospital.

CONCLUSION

This study confirms an increased risk of congenital heart disease among infants with major GI malformations, and particularly amongst those with recognised syndromes. We advocate that infants with major GI malformations should have an early cardiological assessment which should include echocardiogram prior to gastrointestinal surgery and that infants born with such congenital abnormalities should be screened for syndromes and if necessary referred to the genetic service to

enable their parents to be counselled about the potential risk of recurrence in future pregnancies.

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