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MAIN TOPIC

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Additional congenital anomalies in babies with gut atresia or stenosis: when to investigate, and which investigation

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Abstract A wide variety of additional congenital anomalies occur in babies born with a gut atresia or stenosis. The specific pattern of anomalies depends on the location of the atresia. The serious nature of many of them makes perioperative diagnosis imperative. Eighty-six babies born with pure oesophageal atresia (OA), duodenal atresia (DA) or stenosis, or jejuno-ileal atresia (JIA) have been studied. These, combined with over 2,000 cases in the literature, have been used to develop a protocol to optimally investigate babies with gut atresia for associated anomalies. The authors recommend routinely obtaining anterio-posterior and lateral chest and abdominal radiographs for babies with pure OA, DA and intestinal atresia, making sure the entire spine can be visualised. Cardiac and renal ultrasonography (US) should be routine in all babies with pure OA or DA. A micturating cystourethrogram should be done in those babies with abnormal urinary tract US or an associated anorectal anomaly. A sweat test should be obtained in babies with JIA, and a rectal biopsy should be taken in babies with the combination of Down's syndrome and DA to exclude Hirschsprung's disease.

Key words Oesophageal atresia · Duodenal atresia · Intestinal atresia · Congenital anomalies

Introduction

The incidence of additional congenital anomalies in babies born with gut atresia or stenosis is high [8, 10, 11]. The anomalies vary depending on the location of the atresia. The genesis of jejuno-ileal atresia (JIA) is in the late second or third trimester after major organogenesis has occurred. This results in fewer and often less severe associated anomalies than in babies with oesophageal (OA) or duodenal atresia (DA), whose aetiological insult occurs in the first trimester at the time of major organogenesis. The aim of this study was to record the relative frequencies of associated anomalies in babies with different atresias, and to determine for which infants and of which organ systems routine screening investigations are indicated.

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Materials and methods

The study was a retrospective review of the case notes of 86 babies with gut atresia or stenosis treated in the Auckland Children's Hospital between 1979 and 1995. Babies with OA with a tracheo-oesophageal fistula (TOF) and babies with an isolated anorectal malformation (ARM) were excluded from the study group unless they occurred along with DA or intestinal atresia.

Babies were grouped into those with OA without a TOF (n = 10), DA or stenosis (DS) (n = 41), and JIA (n = 35). All associated congenital anomalies were recorded. A literature review was also undertaken to find other large series of babies with atresias to compare our results with and possibly support our recommendations.

Results and discussion

Pure oesophageal atresia

Eight of the 10 babies born with OA or stenosis without a TOF had other anomalies. One who did not have any other anomalies had double perforate webs of the oesophagus (Kluth type VIII [15]). The other 9 babies had a long gap between segments (8 type II 1 and 1 type I 1). There are very few published series dealing with pure OA as a separate group, and almost all of these are small because one hospital will deal with at most one or two cases per year. In 1994, Ein and Shandling from Toronto published a review of 69 babies born with pure OA over a 50-year period [10]. The

Table 1 Cardiac anomalies associated with gut atresia or stenosis

	Oesophageal (n = 10)	Duodenal (n = 41)	Jejuno-ileal (n = 35)
Atrio-ventricular canal defect	0	3	0
Atrial septal defect	2	4	1
Ventricular septal defect	0	0	1
Hypoplastic left heart	0	0	1
Tetralogy of Fallot	0	2	0
Pulmonary stenosis	1	1	0
Patent ductus arteriosus	0	8	2
Aberrant subclavian artery	0	1	0
Persistent left-sided vena cava	1	1	0
Dextrocardia	1	1	0
Total patients (%) with cardiac anomalies	4 (40)	16 (39)	4 (11)

Royal Children's Hospital, Melbourne, described 37 babies with pure OA over a 40-year period [4]. These two series were compared with our own.

Cardiac and great-vessel anomalies were present in 4 babies (Table 1). The Toronto and Melbourne series showed 15% and 21% incidences of cardiac anomalies, respectively [4, 10]. These figures reinforce the recommendation that pre-operative cardiac ultrasonography (US) should be performed in all babies born with OA [18]. Rarely is reparative cardiac surgery necessary prior to initial surgery for OA [18], but it is imperative to know the cardiac status beforehand to optimally manage circulatory problems during or after anaesthesia. The prior knowledge of a right-sided aortic arch is also important, for in this situation an operative approach through the left chest is easier. All babies with suspected OA should have combined anterio-posterior (A-P) and lateral X-ray films of the chest and abdomen. The chest radiograph will supplement the cardiac US giving valuable information on the lung fields and spine, and will also give some information about the length of the upper pouch. The abdominal films will show the rest of the spine and also determine whether or not a distal fistula is present.

Urinary tract anomalies (UTA) were present in 2 of our babies (Table 2), including 1 with bilateral renal dysplasia. The Toronto and Melbourne series had 7% and 16% incidences, respectively [4, 10]. This would suggest that perioperative urinary tract US should be performed, although it does not need to be preoperative unless the baby has not passed urine. The finding of bilateral renal agenesis or bilateral multicystic dysplasia should be confirmed by nuclear renal scan, and the absence of functioning renal tissue should contraindicate surgery. Beasley et al. in Melbourne have recommended routinely performing urinary tract US and a micturating cystourethrogram (MCU) during the initial admission for all babies with OA [5]. Although they did not separate the babies into those with or without a TOF, their overall incidence of UTA was 24%, with a 6% incidence of vesico-ureteric reflux (VUR). They felt that the true incidence of VUR was probably higher, as fewer than 10% of their cases had an MCU performed. The true incidence of VUR is still unknown, but Lander and Drake argue that a routine MCU should be reserved for babies with OA with associated ARM, because only then is the frequency of UTA significantly increased [16]. In both of our cases the UTA would have been detected on US. We currently perform urinary tract US on all babies with OA, and perform an MCU if this is abnormal or if there is an associated ARM. To perform an MCU in all babies with OA is still controversial, and we await the results from centres that do have such a policy.

Three babies had other gastrointestinal (GI) tract anomalies (Table 3), with 2 having ARM. One required a laparotomy for segmental dilatation of the ileum (giant Meckel's diverticulum). Babies with OA do not routinely have an exploratory laparotomy carried out at the time of initial abdominal surgery (usually fashioning of a gastrostomy). There should therefore be a high

Table 2 Urinary tract anomalies associated with gut atresia or stenosis

	Oesophageal (n = 10)	Duodenal (n = 41)	Jejuno-ileal (n = 35)
Bilateral renal dysplasia	1	0	0
Unilateral renal dysplasia	0	1	2
Unilateral renal agenesis	1	1	0
Duplex ureters	0	0	1
Vesico-ureteric reflux	1	3	2
Urethral valves	0	1	0
Bilateral hydronephrosis and hydroureters (unknown cause)	0	0	1
Total patients (%) with urinary tract anomalies	2 (20)	5 (12)	5 (14)

Table 3 Gastrointestinal tract (GIT) anomalies associated with gut atresia or stenosis

	Oesophageal (n = 10)	Duodenal (n = 41)	Jejuno-ileal (n = 35)
Oesophageal atresia with tracheo-oesophageal fistula	–	6	1
Duodenal stenosis	0	–	2
Anorectal malformation	2	5	1
Segmental dilatation of ileum	1	0	0
Hirschsprung's disease	0	1	0
Malrotation	1	15	8
Annular pancreas	0	5	1
Meckel's diverticulum	0	1	1
Intestinal duplication	0	1	1
Gastroschisis	0	1	0
Short-bowel and malrotation syndrome	0	1	0
Pyloric stenosis	0	1	0
Pyloric mass	0	2	0
Large caudate lobe of liver	0	0	1
Total patients (%) with GIT anomaly	3 (30)	24 (59)	9 (26)

index of suspicion of other gut anomalies if there is any unexpected problem with subsequent feeding. The small, disused stomach associated with pure OA is well-documented [22] and should not be classified as a separate anomaly, but one that arises due to failure of the fetus to swallow amniotic fluid [3, 19].

Five babies had orthopaedic malformations, including 2 with lumbosacral anomalies (Table 4). All babies with OA should have full-length A-P and lateral spinal radiographs. The entire spine should be seen on the combined chest and abdominal films taken preoperatively.

Despite a report that tracheomalacia does not occur in babies with pure OA [23], we found 2 babies with clinically significant tracheomalacia and a further 1 with abnormal bronchial anatomy (Table 5). This is consistent with the findings of other studies [17]. We routinely bronchoscope all babies with OA. A proximal TOF, which is present in one-fourth of all babies with OA and a gasless stomach [4], can be excluded by this procedure. This is important, as the presence of a proximal fistula usually means that an immediate repair is possible, the converse holding true in cases of pure OA, where the gap is usually too large.

In summary, babies with pure OA should be investigated in the same way as those with OA with TOF, that is, with cardiac and urinary tract US as well as plain A-P and lateral combined chest and abdominal films. Until the true incidence of VUR has been established from trials, we feel an MCU should be routine only in those babies with abnormal US or an associated ARM (Table 6).

Duodenal atresia and stenosis

Forty-one babies had a DA or DS; 32 (78%) had at least one other anomaly. The most common was malrotation of the bowel (37%), the second most common Down's syndrome (29%). Sixteen (39%) babies had a cardiac or great-vessel anomaly (Table 1); a patent ductus arteriosus (PDA) was the most common (20%). The median gestational age of those with a PDA was 37 weeks, and thus, the PDA did not reflect gross prematurity. Three babies had a atrio-ventricular canal defect, all associated with Down's syndrome. Two babies had tetralogy of Fallot; neither had Down's syndrome. Six studies in the literature with a total of 887 babies with DA or DS reported an overall incidence of cardiac anomalies of 22% (range 17%–34%) [1, 2, 6, 11–13]. As with OA, pre-operative cardiac US is recommended for all babies with DA or DS. A pre-operative chest

Table 4 Orthopaedic anomalies associated with gut atresias or stenosis

	Oesophageal (n = 10)	Duodenal (n = 41)	Jejuno-ileal (n = 35)
Femoral head agenesis	1	0	0
Unstable hips	1	1	0
Talipes equinovarus	0	0	1
Hemivertibrae with kyphoscoliosis	0	0	1
Lumbosacral anomaly	2	2	1
Digit/limb anomalies	2	3	4
Rib anomalies	0	2	3
Total patients (%) with orthopaedic anomaly	5 (50)	6 (15)	5 (14)

Table 5 Miscellaneous anomalies associated with gut atresia or stenosis

	Oesophageal (n = 10)	Duodenal (n = 41)	Jejuno-ileal (n = 35)
Down's syndrome	1	12	1
Other major chromosomal disorder	0	1	0
William syndrome	0	1	0
Cystic fibrosis	0	0	6
Amniotic band sequence	0	0	1
Inguinal hernia/hydrocoele	1	1	2
Undescended testis	2	4	2
Hypospadias/intersex	0	0	2
Russel Silver syndrome	0	0	2
Marfanoid features	0	0	1
Tracheomalacia	2	2	0
Abnormal bronchial anatomy	1	0	0
Cleft lip/palate	0	0	1
Congenital toxoplasmosis	0	1	0
Congenital cataracts	0	1	0
Sacral pit	0	1	0
Single umbilical artery	1	3	1
Dysmorphic face	0	4	1
Congenital ptosis	0	1	0
Squint	0	1	0
Congenital deafness	0	0	1
Preauricular sinus	0	0	1
Rhesus disease of newborn	1	0	0
Fanconi anaemia	1	0	0
Proprionic acidaemia	0	1	0
Total patients (%) with miscellaneous anomalies	6 (60)	25 (61)	14 (40)

radiograph will supplement the cardiac US in determining cardio-respiratory status.

Five (12%) babies had UTA (Table 2); 3 of these had an ARM, and the other 2 had their anomalies detected on US. In previous studies the overall incidence of UTA was 5% (range 2%–16%) [1, 2, 6, 11–13]. However, most of these studies considered only renal anomalies. We feel it would be valuable to perform perioperative urinary tract US in all babies with DA or stenosis and, as with babies with OA, follow up with an MCU if there is an associated ARM or positive US findings.

Other GI anomalies were very common in the presence of DA or DS (59%) (Table 3). Excluding OA with TOF and ARM, 53% of babies with DA or DS had other GI anomalies. This reinforces the recommendation that a full laparotomy should always be performed at the time of definitive surgery. A malrotation was found in 15 cases (37%). The significance of this lesion in the genesis of DA and DS had been discussed previously [14]. Several reports of Hirschsprung's disease (HD) associated with DA have appeared in the literature, with an incidence of 1%–3%. All occurred in the presence of Down's syndrome [2, 6, 11, 25]. The 1 case in our study also had Down's syndrome. This would put the incidence of HD in babies with DA and Down's syndrome at 3%–10%. A policy of selective rectal biopsy in babies with Down's syndrome and DA therefore seems worthwhile.

Six (15%) babies with DA or DS had orthopaedic anomalies (Table 4). Only 2 had a spinal anomaly; 1 had the VATER association and the other Down's syndrome. Irving found an incidence of spinal anomalies in DA or DS of 3%, and all these cases had an associated OA and/or an ARM, both which are associated with a high incidence of spinal anomalies [13]. As with babies born with

Table 6 Recommended routine investigations in babies with gut atresia or stenosis

	Oesophageal	Duodenal	Jejuno-ileal
Cardiac ultrasound	Yes	Yes	No
Renal ultrasound	Yes[a]	Yes[a]	No[b]
A-P and lateral x-rays of chest and abdomen[c]	Yes	Yes	Yes
Sweat test	No	No	Yes
Rectal biopsy	No	Yes if associated with Down's syndrome	No

[a] Micturating cystourethrography should be done in those with an abnormal ultrasound finding or an associated anorectal anomaly
[b] High incidence warrants our institution performing imaging as a clinical trial
[c] Should enable whole spine to be imaged

OA, babies with DA should have plain A-P and lateral chest and abdominal radiographs, which will reveal any gross vertebral and rib anomalies. Although our numbers are small, there did not seem to be any difference in the distribution of anomalies between the babies with DA ($n = 29$) and those with DS ($n = 12$). This suggests that they should both undergo the same investigations.

In summary, the same recommendations for screening for associated anomalies can be applied to DA or DS as to OA. A rectal biopsy should also be performed to exclude HD in babies with Down's syndrome and DA (Table 6).

Jejuno-ileal atresia

Twenty-one (60%) of the 35 babies born with JIA had another anomaly. Four (11%) had a cardiac anomaly (Table 1), 1 of whom had an associated OA with TOF (and so would have merited cardiac US). Two had insignificant septal defects, a patent foramen ovale in one and a small ventricular septal defect in the other. A 3rd baby was cyanosed from birth and was diagnosed as having a hypoplastic left heart, and died aged 8 days without any operation. From four series in the literature covering 736 babies with JIA, only 2% had cardiac or great-vessel anomalies [8, 9, 20, 21]. This suggests that it is not necessary to perform cardiac imaging in babies with JIA unless clinically indicated. Chest and abdominal films should be taken routinely.

Five (14%) babies in our series had a UTA (Table 2). Only 1 of these had associated OA, the other 4 had no other associated major anomaly that would have dictated imaging. Two of the babies had grade III VUR with no other UTA, and thus potentially required an MCU to make the diagnosis. In other series only 1 urinary tract anomaly was identified in 736 babies [8, 9, 20, 21]. The world experience would make it hard to justify routine urinary tract US in all babies with JIA. However, due to the high incidence of UTA in our series we plan to perform urinary tract US for babies with JIA as part of a clinical trial. We understand that this will miss a number of babies with VUR, but feel an MCU cannot be justified at this stage.

Nine (26%) of the babies had other GI anomalies, not including cystic fibrosis (Table 3). Thus, a full laparotomy is essential at the time of surgery. Although we had no babies JIA and HD, this has been reported in one large series with an incidence of 1% [8], and Curry et al. have shown an association with colonic atresia [7]. Cystic fibrosis was present in 6 cases (17%). There was no correlation between the site or type of the atresia in the small bowel and the presence of cystic fibrosis, and therefore, we cannot recommend a selective approach for screening. A sweat test (or blood for cytogenetics) should be performed in all cases of JIA, as the incidence of cystic fibrosis has been constantly reported at around 10% [8, 9, 20, 21, 24].

There was only 1 baby in this group with a spinal anomaly (Table 4), which was obvious clinically as a kyphoscoliosis. This is consistent with the combined reported incidence of 0.3% [8, 9, 20, 21]. As with DA, the preoperative chest and abdominal radiographs will show any gross vertebral anomaly.

In summary, a sweat test and plain abdominal and chest radiographs are the only routine investigations we would recommend to investigate for associated anomalies in babies born with JIA or colonic atresia (Table 6).

Conclusion

The distribution of associated anomalies in babies born with gut atresia or stenosis varies greatly depending on the region of atresia or stenosis. Table 6 summarises our recommendations for investigation of babies with gut atresia.

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