

consultation should be considered. Treatment with tranquilisers brought about an improvement in some patients, including 1 in our study (Table). The use of tranquilisers should, however, be temporary only and these should not be used as a substitute for psychotherapy.

The physician's awareness of the possibility of psychogenic factors in the aetiology of persistent cough may help in early diagnosis and thus repeated troublesome, unnecessary, and expensive investigations may be avoided and the proper treatment be undertaken sooner.

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Cardiovascular anomalies with imperforate anus

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SUMMARY In 68 patients with anorectal malformations cardiovascular anomalies (CVA) were seen in 15 and genitourinary (GU) anomalies in 30. CVA were more frequent (33%) whenever there was a GU anomaly. Ventricular septal defect was the most frequent lesion. All but 1 CVA occurred with type III anorectal malformation. The complexity of the cardiac lesion did not parallel that of the GU anomaly.

The incidence of imperforate anus (IA) has ranged from 1 in 1500 to 1 in 5000 births¹ and cardiovascular anomalies (CVA) may be present in 8-17% of these.^{2,3} In 1 report only, however, were the cardiac lesions outlined.³ Our study was undertaken to determine the type of CVA associated with IA and to establish any correlation between CVA and (i) the anatomic type of anorectal anomaly, (ii) the presence of additional anomalies, especially of the genitourinary (GU) system, and (iii) the degree of complexity of the GU anomaly.

Patients and methods

The records of all 68 patients with anorectal anomalies seen at our hospital from 1974 to 1982 were reviewed. Forty nine infants were referred because

of anal anomaly and the remainder because of multiple abnormalities. Twenty three had been seen previously elsewhere. The diagnosis was made in the newborn period in 66 patients. The initial assessment consisted of physical examination, radiographs, and other tests as required. All patients but 2 with any possibly abnormal cardiovascular findings were seen by a paediatric cardiologist. The anorectal anomalies were classified according to Ladd and Gross—type I, anal stenosis and partial obstruction; type II, imperforate anal membrane; type III, imperforate anus and rectum ending as a blind pouch at variable distance from the skin; type IV, normal anal canal, sphincter, and lower rectum, but upper rectum ending blindly some distance above.¹

Results

Twelve patients had type I anorectal anomaly, 1 had type II, and the remainder had type III. There were 43 boys and 25 girls. The associated anomalies are presented in the Table. There were 15 (12 boys, 3 girls) with CVA. GU anomalies were present in 30 infants, and 10 of them also had a CVA. Of the remaining 38 patients, only 5 had a CVA ($P < 0.05$, χ^2).

Ventricular septal defect, seen in 6 patients, was the most common cardiac lesion. Two premature

Table *Cardiovascular and other anomalies in 68 patients with imperforate anus*

Associated anomaly or syndrome	No	Cardiac lesion	Outcome	
			Alive	Dead
None	28	Ventricular septal defect (1) Aortopulmonary window (1)	28	
Genitourinary: isolated	17	Ventricular septal defect (1) Tricuspid atresia, ventricular septal defect (1)	17	
Genitourinary and spina bifida or meningocele: with tethered cord and club foot (1 with balanced reciprocal translocation involving chromosomes 15 and 4); Sacral dysgenesis	8		7	
	2	Aortic stenosis (1)		
	6	Ventricular septal defect (2) Bicuspid aortic valve (1)		1
Multiple: ambiguous genitalia, double uterus, eventration of diaphragm	1	Patent ductus arteriosus (1)		1
Syndromes:	4		4	
Klinefelter (47,XXY)	1			
Duane with undescended testes	1			
Aarskog	1			
Male phenotype (46,XX)	1			
Polysplenia with situs solitus, undescended testis	1	Pulmonary atresia, atrioventricular septal defect, absence of superior vena cava, left superior vena cava draining into left atrium, coronary sinus ostial atresia, normal pulmonary venous connection (1)		1
Complex: Potter syndrome, hypoplastic lungs, tracheoesophageal fistula: with asplenia, situs solitus, bilateral lobed lung and dysmorphic lower limbs:	2			
	1	Dextrocardia, left superior vena cava draining into coronary sinus, absent innominate vein, normal pulmonary venous connection, agenesis of iliac arteries, abdominal aorta ending in a single umbilical artery (1)		1
with intestinal malrotation, persistent urachus, agenesis of left testis and right leg amelia	1	Patent ductus arteriosus (1)		1
Gastrointestinal:	3		3	
Pyloric stenosis	1	Double inlet ventricle, ventricular arterial discordance, right aortic arch, subpulmonary stenosis, interrupted inferior vena cava and azygos vein continuation and anomalous pulmonary venous connection (1)		
Situs inversus	1			
Tracheoesophageal fistula	1			
Musculoskeletal:	4		4	
Spinal	2	Ventricular septal defect (2)		
Syndactyly	1			
Bifid thumb	1			
Total	68		15	63
			5	

infants with a patent ductus arteriosus were included. Each of the other 7 had different lesions: double inlet ventricle, ventricular arterial discordance, and subpulmonary stenosis with right aortic arch; complete atrioventricular septal defect and pulmonary atresia; aortic stenosis; aortopulmonary window; bicuspid aortic valve; tricuspid atresia and ventricular septal defect; and dextrocardia, agenesis of the iliac arteries and descending aorta ending in a single umbilical artery.

The CVA was confirmed by cardiac catheterisation in 6 infants and by autopsy in 5. CVA occurred in 14 patients with type III anomaly and the exception, a patient with type I anomaly, had a ventricular septal defect. Five of the 68 patients died because of the associated malformations (Table).

Discussion

The 22% incidence of CVA exceeds the expected rate of approximately 1% of live births with congenital heart defects. In a previous report tetralogy of Fallot was the most common lesion and ventricular septal defect the second most common.³ In our study tetralogy of Fallot was not seen. Features of Ivemark syndrome in which IA is uncommonly present were seen in 3 patients. The incidence of CVA was greater in the presence of a GU anomaly.

Anorectal anomalies may occur isolated or in patterns of congenital malformations such as the VACTERL association,^{4,5} caudal regression syndrome,⁷ or vertebral agenesis⁷ and in these cardiovascular malformations are often seen. A new

syndrome with IA, hypothalamic hamartoblastoma, and other anomalies including GU and cardiac has been described by Hall *et al.*⁸ Noonan-Suldino, 'C', Kaufman, cat eye, and Meckel syndromes may also have IA and CVA.⁸ Anorectal anomalies may be component of any inherited or chromosomal abnormality or may be caused by an environmental agent.⁴

The link between the cardiovascular system and the caudal area is unknown. The embryologic development of the urinary, genital, and rectal areas occur between the 4th and 8th weeks of gestation when the cardiovascular system is also being formed. Anorectal and GU malformations are caused by arrests and abnormalities of the embryonic development of the caudal region.⁶ Alteration of the mesodermal cell migration during the primitive streak period has been proposed to explain disparate defects of the cranial and caudal ends. Teratogens implicated in anorectal and GU anomalies have included thalidomide, progesterone (alone or with oestrogen), and maternal diabetes.⁴ It is possible that in some instances the same factor may cause simultaneously malformations of the caudal region and of the cardiovascular system. In our study the most serious CVA occurred in the absence of important GU malformations.

The importance of the anorectal anomaly may be secondary when compared with the associated malformations. The mortality rate in IA has been 8–27% and in most instances death is due to the associated anomalies.

The high incidence and the severity of associated malformations in type III anorectal anomaly warrant a full assessment before any attempt to correct or palliate the anorectal malformation. Its

role may be a less important one in view of the seriousness and complexity of the associated malformations, especially of the renal and cardiac systems.

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