CARDIAC ANOMALIES IN MONGOLISM

BY

PHILIP RAINSFORD EVANS

From the Hospital for Sick Children, Great Ormond Street, London

Received November 23, 1949.

In a well-known textbook Maud Abbott is credited with the opinion that " the commonest cardiac anomaly in Mongolian idiocy is a defect of the interauricular septum, especially ostium primum." Reference to her paper shows that she actually wrote that in mongols the defect " in our experience is not infrequently of this type " (Abbott, 1924). She described one case, in which there was also a ventricular septal defect that divided the aortic cusp of the mitral valve.

This addition is suggestive of persistent atrioventricularis communis, of which Taussig (1947) wrote: " The diagnosis is based on the law of probability. If a Mongolian idiot has a congenital malformation of the heart which superficially resembles a ventricular septal defect and there is no cyanosis, the overwhelming probability is that he has a patent atrioventricularis communis. The demonstration of an abnormally low oxygen saturation of the arterial blood clinches the diagnosis." Robson (1931), Meecker (1935) and Robinson (1941), have described examples. Goetsch's patient showed the same lesion and dextroposition and infantile coarctation of the aorta (Goetsch, 1938), while two patients described by Gunn and Dieckmann (1927) showed mongolism, persistent atrioventricularis communis and patent foramen ovale, with patent ductus arteriosus also in one case.

One might easily get the impression that persistent atrioventricularis communis was the usual cardiac lesion of mongolism, in spite of the description of the single cases with other anomalies by Wolf and Levinson (1939) and Mannheimer (1949). However, Leech (1935) described six cases, only one of which had this defect, with patent foramen ovale and anomalies of the great vessels in addition. Writers on mongolism (e.g. Benda, 1946, and Engler, 1949) do not make numerical statements about the frequency of the lesions they mention. The examination of a larger series than that of Leech would seem to be worth while.

Between January, 1911, and October, 1949, necropsies were done at the Hospital for Sick Children, Great Ormond Street, on 63 mongols, and in 28, cardiac anomalies were described. The records are not impeccable, for until 1942 necropsies were not made by a trained morbid anatomist, but by the Medical Registrar and Pathologist, who was selected for the three years' appointment because of clinical competence and ability. The diagnosis of mongolism is likely to have been right in every case, but the descriptions of the cardiac lesions must be viewed critically. In many cases they are excellent. However, in analysing the findings, " probe," " physiological " or similar slight degrees of patency of the foramen ovale at all ages, and all but wide patencies of the ductus under the age of one year have been omitted. " Patent foramen ovale " has not been taken at its face value, as in earlier years there may have been confusion with persistent ostium primum.

In the period reviewed, children under 12 years of age, suffering from all types of acute disease (except infectious fevers) and many chronic medical and surgical conditions were admitted. Judging by the clinical summaries, congenital heart disease was the major cause of death in seven mongols, broncho-pneumonia in another nine, and extracardiac congenital anomalies in three. The age at death was rather similar in mongols and other children with congenital heart disease (Table I). Nine of the mongols with heart disease showed other congenital anomalies: Meckel's diverticulum

TABLE I

AGE AT DEATH IN MONGOL IMBECILES WITH AND WITHOUT CONGENITAL HEART DISEASE, AND IN OTHER CHILDREN WITH CONGENITAL HEART DISEASE

Death in	Mongol		Other children with morbus cordis
	With morbus cordis	Without morbus cordis	
1st month	4	5	6
2nd–5th month ..	16	7	35
6th–11th ,, ..	6	7	5
2nd year	1	7	2
3rd ,, ...	0	4	0
4th ,, ...	0	4	0
5th ,, ...	1	0	1
6th ,, ...	0	1	0
7th ,, ...	0	0	1

in three, abnormal pulmonary fissures in two, atresia of the common bile duct, bifid rib, cleft palate, symphalangy, œsophageal atresia with œsophagotracheal fistula, hydrocephalus, spina bifida with meningocele, ectopic anus, duodenal atresia, and intestinal malrotation, each in one instance.

The anomalies found in these 28 hearts are surprisingly various (Table II). Interestingly enough, persistent atrioventricularis communis was found only 4 times, and even allowing for unfamiliarity with the condition, it must have been absent in at least 21. On the evidence produced by these 28 and Leech's 6 cases, persistent atrioventricularis communis should not be regarded as the typical cardiac lesion of mongolism, for it occurred in only 5 of the total of 34. Application of the " law of probability " mentioned by Taussig leads to the conclusion that " if a Mongolian idiot has a congenital malformation of the heart which superficially resembles a ventricular septal defect and there is no cyanosis," the probability is that he actually has a ventricular septal defect, although he may have the less common persistent atrioventricularis communis.

TABLE II

CARDIAC ABNORMALITIES FOUND IN 28 MONGOLS

	Without patent ductus arteriosus	With patent ductus arteriosus
Large atrial septal defect, type uncertain	5	—
Persistent ostium primum	1	—
Persistent ostium primum and secundum	—	1
Absent auricular septum	1	—
Persistent atrioventricularis communis	3	1
Large atrial septal defect, and ventricular septal defect (membranous)	1	2
Ventricular septal defect (membranous 6, more extensive 1)..	6	1
Patent ductus arteriosus	—	1
Pulmonary infundibular stenosis	1	—
Pulmonary artery stenosis, atrial septal defect ..	—	1
Fallot's tetralogy (1 with atrial septal defect) ..	2	—
Aberrant right subclavian artery	—	1

In the 28 hearts there were 47 anomalies of the heart or great vessels (Table III). Atrial and ventricular septal defects each accounted for just over one-third, and patent ductus arteriosus for one-sixth. Most of the remaining lesions were pulmonary stenoses. Defect of the pars membranacea of the ventricular septum is the commonest anomaly. Owing to uncertainty about the

TABLE III

FREQUENCIES OF INDIVIDUAL ANOMALIES IN THE HEARTS OF 28 MONGOLS WITH CONGENITAL HEART DISEASE

Auricular septum, all defects		17
,, ,, absent	1	
,, ,, persistent ostium primum	2	
,, ,, ,, secundum	1	
,, ,, ,, atrioventricularis communis	4	
Ventricular septum, all defects		17
,, ,, persistent atrioventricularis communis	4	
,, ,, membranous defect	12	
,, ,, absence of four-fifths	1	
Ductus arteriosus, widely or persistently patent		8
Pulmonary stenosis		4
,, ,, infundibular	1	
,, ,, arterial	1	
,, ,, infundibulo-arterial	2	
Aberrant right subclavian artery		1

precise nature of the atrial septal defects it is impossible to decide whether persistent ostium primum is a very close runner-up, or merely a bad second. Probably patent ductus comes third, while pulmonary stenosis and persistent atrioventricularis communis are bracketed in the fourth place. Other lesions were inconsiderable in this series.

This examination of records showed that no particular combination of anomalies was characteristic of the abnormal heart in mongolism, but it did not indicate whether the uncommon lesion of persistent atrioventricularis communis was in fact a peculiarity of the disease.

The necropsy records of fifty children, who were not mongols but who had congenital anomalies of the heart were therefore examined. They were chosen from the same twenty volumes of post-mortem reports as the mongols, the first two or three cases (depending on whether there were more or less than the average number of mongols in that particular volume) being selected from each. The list (Table IV) demonstrates the amazing variety of congenital heart disease in patients dying in a children's hospital.

Among the individual lesions (Table V), the absence of persistent atrioventricularis communis is noteworthy: it appears to be less rare in mongols than in other children with congenital heart disease. Auricular septal defect in general is probably unduly common in mongols, while ventricular septal defect and patent ductus are not. On the other hand, transposition of the great vessels seems to be unusual in mongolism, although Mannheimer's case must not be forgotten (Mannheimer, 1949). The non-occurrence of truncus arteriosus and of aortic coarctation in the mongols is notable, although the series is small. The adult type of coarctation is not likely to be encountered often in the autopsy records of a children's hospital, but clinical experience also indicates that it is uncommon in mongols. The relatively higher figure for pulmonary stenosis in the mongols is rather surprising. It is, of course, possible that the prognosis of mongols with pulmonary stenosis, atrial septal defect, or persistent atrioventricularis communis is worse than that of other patients with these cardiac anomalies, who may come to necropsy in a hospital for adults. The figures presented here show the incidence of different congenital abnormalities of the heart in patients dying young, rather than in all patients.

The most clear-cut difference between the lesions in the two series is the absence of defects due to malrotation of the bulbus cordis (except Fallot's tetralogy) in the mongols, and of a common atrioventricular opening in the other children. In this series atrial septal defects (which may have been due to persistence of the ostium primum) were commoner in the mongols than in the other

TABLE IV

CARDIAC ABNORMALITIES FOUND IN 50 PATIENTS WHO WERE NOT MONGOLS

	Without patent ductus arteriosus	With patent ductus arteriosus
Atrial septal defect (1 certainly ostium primum)	3	1
Ventricular septal defect	5	3
Patent ductus arteriosus	—	7
Atrial and ventricular septal defects	1	1
Bilocular heart, subaortic atresia	—	1
Bilocular heart, severe pulmonary artery stenosis, single pulmonary vein	1	—
Trilocular biatrial heart, atrial septal defect	1	—
Trilocular biventricular heart	1	—
Trilocular biventricular heart, atrial septal defect (1 with mitral atresia)	1	1
Hypoplastic left auricle and ventricle	1	1
Tricuspid atresia with non-functioning right ventricle, atrial and ventricular septal defects	1	—
Truncus arteriosus, over-riding interventricular septum (1 with atrial septal defect)	2	—
Truncus arteriosus from right ventricle, ventricular septal defect	2	—
Transposition of aorta and pulmonary artery (1 in each group with atrial septal defect and 1 in each group with V.S.D.)	4	1
Transposition of aorta, pulmonary artery over-riding ventricular septum	1	—
Pulmonary artery stenosis, atrial septal defect, ventricular septal defect	1	—
Pulmonary artery stenosis, ventricular septal defect	—	1
Coarctation of aorta, infantile type	—	2
Coarctation of aorta, adult type (1 with ventricular septal defect)	2	—
Situs inversus of heart alone (1 with atrial and 1 with ventricular septal defect)	2	—
General hypertrophy	1	—
	50	

TABLE V

INDIVIDUAL ANOMALIES OCCURRING 2 OR MORE TIMES IN THE HEARTS OF 28 MONGOLS, OR 4 OR MORE TIMES IN 50 PATIENTS WITH CONGENITAL HEART DISEASE WHO WERE NOT MONGOLS

Lesion	Mongol		Not mongol	
	No.	Percentage	No.	Percentage
Auricular septum, all defects	17	60	18	36
Auricular septum, ostium primum or secundum, without persistent atrioventricularis communis	12	43	15	30
Persistent atrioventricularis communis	4	14	0	0
Ventricular septum, all defects	17	60	26	52
Ventricular septum, absent or nearly so	1	4	5	10
Ventricular septum, membranous defect without persistent atrioventricularis communis	12	43	21	42
Patent ductus arteriosus	8	29	20	40
Truncus arteriosus	0	0	4	8
Pulmonary stenosis	4	14	3	6
Coarctation of aorta	0	0	4	8
Transposition of aorta and pulmonary artery	0	0	7	14

children, ventricular septal defects were equally common in the two groups, while patent ductus arteriosus was less common in the mongols. The common atrioventricular canal is divided and the ostium primum is closed at about the 41st day of intrauterine life; the interventricular foramen closes at about the 47th day (Hamilton, Boyd, and Mossman, 1945); the second torsion of the bulbus occurs between the fifth and eighth weeks (Brown, 1939) so that exaggerated torsion may occur after the 56th day; the ductus arteriosus closes after birth. Comparison of this time table with the lesions found in or absent from the mongol series supports Abbott's suggestion that the defects of development in mongols occur early (Abbott, 1924).

SUMMARY

Congenital heart disease was found in nearly half of a series of mongol imbeciles dying in the first five years of life. In the records of 63 autopsies, 28 examples were found.

Ventricular and atrial septal defects each accounted for one-third of the individual lesions, and patent ductus arteriosus for one-sixth. Pulmonary stenosis and persistent atrioventricularis communis each occurred in 4 patients.

Compared with other children with congenital heart disease dying in the same hospital in the same period, transposition of the great vessels, coarctation, and truncus arteriosus seemed to be unassociated with mongolism. Atrial septal defects and pulmonary stenosis were more common in the mongols than in the other patients. Speaking generally, the cardiac anomalies in mongolism appeared to be due predominantly to defects occurring in the earlier stages of cardiac development.

My thanks are due to Dr. Maurice Campbell for his interest in the preparation of this paper.

REFERENCES

Abbott, M. (1924). *Bull. Int. Assn. Med. Mus.*, **10**, 111.
Benda, C. E. (1946). *Mongolism and Cretinism*, New York.
Brown, J. W. (1939). *Congenital Heart Disease*, London.
Engler, M. (1949). *Mongolism*, Bristol, 48.
Goetsch, C. (1938). *J. Tech. Meth.*, **18**, 117.
Gunn, F. D., and Dieckmann, J. M. (1927). *Amer. J. Path.*, **3**, 595.
Hamilton, W. J., Boyd, J. D., and Mossman, H. W. (1945). *Human Embryology*, Cambridge.
Leech, C. B. (1935). *J. Pediat.*, **7**, 802.
Meecker, L. M. (1935). *J. Tech. Meth.*, **14**, 72.
Mannheimer, E. (1949). *Morbus Caeruleus*, Basel and New York, 294.
Robinson, D. W. (1941). *Arch. Path.*, **32**, 117.
Robson, G. M. (1931). *Amer. J. Path.*, **7**, 229.
Taussig, H. B. (1947). *Congenital Malformations of the Heart*, New York.
Wolf, I. J., and Levinson, S. A. (1939). *Amer. Heart J.*, **18**, 241.