AGENESIS OF LUNG

BY

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Agenesis of a lung is a rare lesion. Reviews of all reported cases have been published by Hurwitz and Stephens (1937); Deweese and Howard (1944); Smart (1946); Per Wexels (1951); Oyamada, Gasul, and Holinger (1953), and Valle (1955). The last author collected and tabulated details of 120 cases. Since Valle's (1955) publication, cases have been reported by Warner, Palladino, Schwartz, and Schuster (1955); Clark, Scott, and Johnson (1955); Hochberg and Naclerio (1955); Levy (1955); Bariety, Choubrac, Vaudour, Tupin, and Manouvrier (1955); Sánchez Barrios and Escobar Aces (1956); Rouco Ajá, Codinach, and Segura (1956); Chambers and Tancredi (1957), and Netterville (1957). A list of references to the early work on the subject will be found in Smart's (1946) paper.

We now report the findings in four proven cases of total agenesis of a lung and discuss certain features of the disease, in particular, some aspects affecting the clinical management of the condition. We have excluded all cases in which the exact diagnosis is in doubt, namely, cases of lobar agenesis, and those in which the reduction in the amount of lung tissue is due to some secondary or acquired cause.

The difficulties in diagnosis during life are considerable. In a number of previously reported cases, confirmation of the congenital origin of the condition by necropsy or thoracotomy is lacking. These variations in criteria for accepting the diagnosis reduce the value of any statistical analysis of reported cases, and have, in part, been responsible for a number of different conclusions being reached about the disease by various authors. Of the 104 cases mentioned by Smart (1946), 29 were considered incorrectly diagnosed, and, of these, 21 were thought to be examples of gross fibrosis or congenital atelectasis. Schneider (1948) described two cases thought to be suffering from agenesis of the right lung in infancy. These two patients were reviewed at intervals over a period of 15 years, and by bronchography were shown to have at least one normal lobe on the side of the supposed absent lung. The original diagnosis was revised. Per Wexels (1951) described a number of case reports suggestive of agenesis: one of these cases was an infant suffering from congenital atelectasis.

In older patients it is a not uncommon experience to find identical radiographic and bronchoscopic appearances to those of agenesis, a result, for instance, of tuberculous stricture of a main bronchus with fibrosis throughout the lung. Usually some fact in the patient's history or feature on examination clarifies the diagnosis. There are patients, however, from whom a history of some acquired cause to account for the clinical findings is unobtainable. From our own experience a case is relevant. The patient, a child of 15, had a routine chest radiograph and appeared to be suffering from total agenesis of the left lung. Bronchoscopy demonstrated a small blind pouch representing the left main bronchus and a bronchogram apparently confirmed the diagnosis of pulmonary agenesis. No previous history was offered, but subsequently it was learned from a relative that the child had been run over by a farm cart when aged 3. A radiograph shortly after the accident demonstrated a shallow pneumothorax but an otherwise normal left lung. The correct diagnosis was one of traumatic stricture of the bronchus. The patient knew nothing of the injury, and we wish merely to make a point of reiterating the uncertainty in reaching a diagnosis of such a condition as pulmonary agenesis.

The diagnostic difficulty arises from a number of reasons. There are no characteristic symptoms or clinical signs of pulmonary agenesis. The chest wall is likely to be normally developed and symmetrical. Schmit (1893) described the necropsy findings in an infant born without either lung. The development of the chest wall was normal, and he concluded that the formation of the thoracic cage was independent of normal lung growth. Other authors (Smart, 1946; Hochberg and Naclerio, 1955) have confirmed the symmetry of the two sides in cases of unilateral agenesis.
AGENESIS OF LUNG

Nevertheless, study of the cases reported in the literature reveals that the physical signs of this disease are too variable for importance to be attached to such manifestations as the shape of the patient's chest.

The signs of associated congenital lesions may exceed those of the lung condition, as in the four cases reported here, and the degree of disability produced may vary from death at birth to negligible symptoms throughout life and death in old age—at 72 years in the case reported by Heerup (1927). A diagnosis based on radiographic, bronchoscopic, and the appearances at bronchography can be presumptive only and offers nothing more tangible than a strong suspicion that agenesis of lung exists. Two ancillary methods can contribute useful information. Tomography may outline bronchial markings beyond an acquired occlusion of a main bronchus. These bronchi remain unfilled at bronchography where the occlusion is complete, and their demonstration by tomography is certain evidence of development of lung tissue to a degree which does not permit of a diagnosis of congenital agenesis.

The other method is angiocardiology. This may be used in all cases of suspected agenesis where the establishment of a diagnosis is indicated; in short, those with symptoms. From the angiocardiological findings much may be deduced about the degree of agenesis and whether its origin is congenital or acquired. Furthermore, information becomes available to indicate the benefit from surgical exploration—a factor in the management of the disease in which clinical practice seems to vary. The interrelationship of absence of a pulmonary artery and agenesis of the lung on the same side cannot be succinctly stated. Where there is no lung tissue of any sort the pulmonary artery on the ipsilateral side is absent. Schneider (1912) stated this in describing the various degrees of agenesis. In Schneider's agenesis, grade I and II, no lung tissue is formed on the affected side, and the main pulmonary artery sends its only branch to the existing lung, an observation which has been many times confirmed. The corollary to this, namely that the presence of branches of the pulmonary artery, however small or ill-formed, to the side of the supposed agenesis is evidence that lung tissue in some stage of maturity exists, must also be true. The reason for the development of the foetal pulmonary artery is the existence of pulmonary tissue for the artery to grow into and vascularize, and although many causes may retard or deform growth of either the lung or the artery, or both, some lung must exist where a pulmonary artery is demonstrable. This simple method of classifying degrees of pulmonary agenesis of the lung. There is no case recorded become obvious during the last decade that the pulmonary artery may be absent even though a well-developed lung is present. The literature on the subject has been reviewed recently by Barthel (1956) and by Emanuel and Pattinson (1956). In none of the cases of aplasia of a pulmonary artery described in these two publications has it coexisted with agenesis of lung, and where the findings suggest a diagnosis of pulmonary agenesis the angiographic demonstration of an absent artery confirms, for all practical purposes, total agenesis is complicated by the fact that it has which makes this supposition untenable, with the possible exception of Case 19 recorded by Jones (1955). In this patient the radiographic appearances were those of lung agenesis. At operation no pulmonary artery was present, and it was found that the lower lobe was supplied by systemic vessels from the aorta and an intercostal artery.

The angiocardiological findings in patients with the picture of agenesis of the lung suggest a classification of the disease which has the sound advantage of offering some indication of the benefit from surgical treatment, an aspect of the disease which we wish to discuss. Classification has been obscured by the number of descriptive terms, such as agenesis, hypogenesis, aplasia, and hypoplasia, used to describe similar conditions, and the different interpretations of Schneider's (1912) original views on the anatomy of the various degrees of agenesis. Particularly in the less severe degrees there is no certain method of differentiating an acquired pulmonary hypoplasia from a lesion of congenital origin. The classification proposed enables the clinician to avoid such issues. Our proposals are that in patients with the radiographic and bronchoscopic appearances of agenesis (a) where angiocardiology demonstrates the pulmonary artery to be absent no pulmonary tissue exists and the condition is true congenital agenesis; and (b) where angiocardiology demonstrates a pulmonary blood flow, some lung must exist and the condition is to be regarded as pulmonary hypoplasia, which may be congenital in origin, or arise from an acquired cause, such as foetal atelectasis, stricture of a bronchus, or infantile diaphragmatic hernia (Roe and Stephens, 1956).

We consider it more important to establish the degree of agenesis by this relatively simple means than to decide whether the defect has congenital
or acquired origin. What lung there is, in the hypoplastic conditions, is likely to be atelectatic, fibrosed, functionless, and prone to infection, whatever the aetiology, and if the patient has symptoms an indication for surgical exploration may exist. Where the angiocardiogram fails to demonstrate a pulmonary artery, that is, in true congenital agenesis, there is no indication for exploration because no lung is present.

Our cases are all examples of congenital agenesis, which, using Schneider's classification, are of grades I and II.

CASE REPORTS

CASE 1.—A boy, admitted to hospital on the fifth day of life, had regurgitated all his feeds since birth. He was normally developed, except for the external ears. These were replaced by shapeless rudimentary pinnae. The right radius was absent. The child died from inhalation bronchopneumonia on the sixth day.

Summary of Necropsy Findings.—The right lung was absent. The right main bronchus was represented by a small blind pouch, surrounded by fat. No pulmonary artery was present on the right side. Apart from pneumonic changes, the left lung was normal. Two veins from the left lung drained into the left auricle. There were no other pulmonary veins. The heart was large and weighed 12.8 g. It was lying against the chest wall in the right axillary line. There was a small patency of the interatrial septum. The upper part of the oesophagus terminated as a blind sac at the level of the sixth tracheal cartilage. It was separated by a narrow area of connective tissue from the lower end of the oesophagus. The trachea and lower segment of the oesophagus communicated. The right radial bone was absent.

CASE 2.—A girl, aged 14 weeks, was admitted to hospital under the care of Dr. Braid. The child was first-born, weighed 7 lb. at birth, and for the first three days of life was nursed in an oxygen tent. On admission it was marasmic (weight 7 lb.). Bilateral accessory auricles of the ears and bilateral inguinal herniae were present. A loud systolic murmur was audible over the praecordium. The child died one week after admission, at the age of 15 weeks. A chest radiograph taken shortly before death is shown (Fig. 1).

Summary of Necropsy Findings.—The left lung was absent. There was no left bronchus, and the trachea, which appeared to have cartilaginous rings around its whole circumference, ran straight into the right lung. There was no left pulmonary artery. The right lung was normal except for terminal bronchopneumonia. The heart was dilated and filled the left pleural cavity. An interventricular septal defect and rudimentary aorta were present. The left kidney was absent. In addition to the accessory ears, the child had a macrostoma and a defect of the bony palate.

CASE 3. A boy, weighing 6½ lb. at birth, aged 9 days, was admitted to hospital under the care of Mr. Baines. An imperforate anus and bilateral absence of the radial bones were found on external examination. A gap of over 1 cm. existed between a skin marker on the anal skin dimple and the gas bubble in the rectum. Absence of the left lung and a congenital heart lesion were also detected. The child died, aged 16 days, from bronchopneumonia.

Summary of Necropsy Findings.—The left lung was absent. The left main bronchus was atrophic. No pulmonary artery to the left side existed. The right lung contained multiple areas of consolidation and its lower lobe projected abnormally toward the mediastinum, otherwise the right lung was normal. An enlarged heart filled the left thoracic cavity. The interventricular septum was patent and the interatrial septum widely so. The wall of the right ventricle was hypertrophied. The aorta originated from both the left and right ventricles. Two superior vena cavae were present. The only abnormality in the alimentary tract was the imperforate anus. The rectum terminated blindly about 1 cm. above the anal dimple. There was no recto-urethral or recto-vesical fistula. The left kidney was hypoplastic and its parenchyma replaced by numerous cysts. Bilateral absence of the radial bones was confirmed.

CASE 4.—A woman, aged 36, first came under observation in 1952. She had no symptoms and physical examination was negative except for some flattening of the left side of the chest and a systolic murmur audible over the praecordium. A chest radiograph is shown (Fig. 2). She was unwilling to submit to any investigation with the object of establishing a diagnosis, and continued to do her normal work for the next four years. Shortly before her death she was admitted to hospital in congestive...
AGENESIS OF LUNG

31

oedematous, but structurally normal. The heart was enlarged and a pericardial effusion was present. There was a defect in the interventricular septum, but no other developmental defect in the thorax or elsewhere in the body.

Table I demonstrates the variety of developmental defects associated with these examples of agenesis of lung. In certain cases an environmental rather than a hereditary factor may influence the foetus and cause the abnormality. This is illustrated by Boyden’s (1955) description of agenesis of the lung in one of identical twins. Of all the associated defects, absence of the radius is one of the most engaging. In the cases reported this defect was present in two; in one patient on the same side as the absent lung, and in the other the radial defect was bilateral. This association has been described previously in a number of reports. The reason for this particular bone in the embryonic fore limb bud being picked out is not clear, nor is the significance of the association. It is possible that at a stage in the development of the foetus the cells forming the radius and those forming the lung lie in proximity and may be affected by the same environmental factor, and if this is the case the combined defect must be established by the very earliest days in the pregnancy. Experimental embryological data on the subject are not available. Riordan (1955) described the treatment of 11 cases of absence of the radius; three of these exhibited the radiological appearances of agenesis of the lung, on the same side. The lesions were right-sided. An explanation of the association of these two rare anomalies could not be offered by the author.

The associated abnormalities may be divided into those causing death, such as oesophagotracheal fistula; those affecting survival, such as atrial septal defects and patency of the ductus arteriosus; and the remainder, which do not affect expectation of life. In the last group are absence of the radius, accessory ears, minor anatomical changes in the existing lung, and a number of other defects inconsequential as far as survival is concerned. Absence of a lung is in itself a defect compatible with an almost normal existence, and the associated defects, oesophagotracheal fistula for instance, govern the prognosis. In these circumstances surgical treatment of a serious associated lesion, particularly when single, should be offered. Nicks (1957) has described ligation of a patent ductus arteriosus in a patient with a total left-sided pulmonary agenesis. The patient, a girl of 7, was transformed into a vigorous child by the operation. Maier and Gould

cardiac failure. The increased congestion in the right lung field is shown (Fig. 3). She died at the age of 40 from congestive cardiac failure.

Summary of Necropsy Findings.—The left lung was absent and the trachea ran to the right lung. There was no pulmonary artery to the left lung. The right pleural cavity contained a large effusion of straw-coloured fluid, and the right lung was congested and
(1953) described tracheal compression due to an abnormality of the left pulmonary artery in a child with agenesis of the right lung. The details of the vascular abnormality, the findings at operation, and the result are reported.

The diagnosis of congenital heart disease in early childhood may not be easy and the propriety of urging surgical correction of an additional defect should be qualified by noting the high incidence of congenital heart disease in the cases reported in this paper and in many of those in the literature.

There are differences in the prognosis between agenesis of the right and left lungs. The average age at death is lower in right-sided lesions and the reasons for this are not clear. An analysis of the total number of cases described by Hurwitz and Stephens (1937), by Deweese and Howard (1944), and by Per Wexels (1951) reveals that the average age at death of patients with agenesis of the right lung is 6 years, whereas that of left-sided lesions is 16.

In a paper on the prognosis of pulmonary agenesis according to the side affected, Schaffer and Rider (1957) pointed out that absence of the right lung carries the worse prognosis. They discussed a number of points which might affect the result in any statistical analysis, and because of a possibility of unknown bias their conclusions were only tentative. They stated that the reason for the worse prognosis in right-sided lesions might lie in the greater shift of the mediastinum, causing compression of the great vessels and trachea when this side is affected, and that on the basis of the collected figures of Oyamada et al. (1953) they could find no greater incidence of associated lesions when the right lung was absent. We should hesitate to accept either of these tentative conclusions. In an analysis of the cases tabulated by Hurwitz and Stephens (1937) and by Per Wexels (1951) associated lesions reducing expectancy of life were present in 10 out of 28 right-sided lesions, and in 10 out of 41 left-sided lesions; there appears to be a greater incidence of serious additional abnormalities when the right lung is absent. Further evidence that this is so is afforded by examination of the reported cases from a different angle. Valle (1955), in the most recent review, collected 120 cases (54 right-sided, 63 left-sided, and three bilateral): of the cases diagnosed during life, 16 were left-sided and five right-sided. In none of these cases could the associated lesion be regarded as serious. We suggest that where the left lung is absent, serious associated defects are less common and that it is for this reason that patients with left-sided agenesis live longer. Valle's (1955) figures also reveal the differences between the two sides in patients first diagnosed after the tenth year of life. When the left lung was absent, 25 patients were diagnosed after the tenth year of life, and only eight where the right lung was agenetic. This is tantamount to stating that serious associated defects occur less frequently on the left side.

Death in each of our patients was attributable to some other defect than agenesis of the lung. The actual cause of death in some of the case reports in the literature has not been specifically mentioned, and any supposition, therefore, that right-sided agenesis is, by itself, a graver lesion is untenable. The suggestion put forward by Schaffer and Rider (1957) concerning the compressing effect of increased shift of the mediastinum, when the right lung is absent, on the great vessels and trachea is one on which definite evidence must necessarily be difficult to collect. Maier and Gould (1953) in their case report clearly demonstrated compression of the trachea by the arterial abnormality at operation, although at necropsy this effect was no longer pronounced. The living pathology differed from that at necropsy.

Any discussion of the place of surgery in the management of pulmonary agenesis might include the possible beneficial effect from a space-reducing operation on the affected side. Although the term "space-reducing" is used descriptively, it is, of course, a misnomer. No space exists, on the side

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**Table 1**

DEVELOPMENTAL DEFECTS IN PRESENT SERIES

<table>
<thead>
<tr>
<th>Sex</th>
<th>Side</th>
<th>Age at Death</th>
<th>Type of Agenesis</th>
<th>Pulmonary Vessels</th>
<th>Other Defects</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>Male</td>
<td>Right</td>
<td>6th day</td>
<td>Schneider's Type II</td>
<td>Absent</td>
</tr>
<tr>
<td>Case 2</td>
<td>Female</td>
<td>Left</td>
<td>15 weeks</td>
<td>..</td>
<td>..</td>
</tr>
<tr>
<td>Case 3</td>
<td>Male</td>
<td>..</td>
<td>16th day</td>
<td>..</td>
<td>..</td>
</tr>
<tr>
<td>Case 4</td>
<td>Female</td>
<td>..</td>
<td>40 years</td>
<td>..</td>
<td>..</td>
</tr>
</tbody>
</table>

**Notes:**
- Interventricular septal defect.
- Imperforate anus.
- Cleft palate.
- Absent left kidney.
AGENESIS OF LUNG

of the agenesis, which in any way resembles the space after pneumonectomy. Schneider (1912) described the anatomical changes which take place during foetal life to compensate for the missing viscera and at birth displacement of the mediastinum, elevation of the diaphragm, a large thymus, and an increase in fat participate in filling the hemithorax.

The theoretical value of a space-reducing operation is twofold: the centralization of the mediastinum to reduce compression of the great vessels, and the prevention of over-distension of the existing lung. As to the first, the evidence in favour seems too nebulous to recommend it, apart from the considerable practical difficulty of defining a satisfactory space-reducing operation in infancy and childhood. Its value in preventing over-distension of the existing lung is also a matter for conjecture. There are two descriptions of full respiratory function studies in children with pulmonary agenesis, those of Warner et al. (1955) and Clark et al. (1955). Neither of these groups of workers were convinced of the need for operation by the results they obtained. It would seem desirable to await the results of investigating the degree and rate of progress of respiratory dysfunction over a number of years before recommending such a procedure in cases of pulmonary agenesis.

As far as operative exploration is concerned, the indications must be based on the angiocardiographic findings in uncomplicated pulmonary agenesis. Where no pulmonary artery is demonstrable, we believe the diagnosis of total pulmonary agenesis to be sufficiently established by this investigation as to make any confirmatory operation unnecessary. In these circumstances surgical exploration can make no additional contribution, either in diagnosis or treatment. If, however, a demonstrable pulmonary artery coexists with the clinical and radiographic findings of pulmonary agenesis, it is certain, as already suggested, that a lung remnant is present and its excision may relieve symptoms. In pulmonary agenesis associated with a serious deformity, the possibilities of surgical correction of the latter are apparent from Maier and Gould's (1953) and Nicks' (1957) case reports.

SUMMARY

Four cases of pulmonary agenesis are reported. Certain features of the disease are discussed.

We should like to thank Dr. H. S. Baar for allowing us access to his complete post-mortem records of the first three cases, and for his permission to publish his findings.

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Agenesis of Lung

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