Congenital Pulmonary Agenesis

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In commenting upon the genesis of the lung, Gruenfeld and Gray state that “developmental anomalies of the lung is favored by the phylogenetic newness of this organ and the enormous growth it must undergo in the embryo before its functional form is achieved.” These authors classify the maldevelopment of the lung as follows:

A—Deficiency in development of the lungs.
1—Absence of both lungs
2—Agenesis of one lung
3—Abnormalities in the branchings of the stem bronchi
4—Cystic disease and so-called congenital bronchiectasis.

B—Excessive formations in the development of the lungs.
1—Azygos lobe
2—Supernumerary lobes
   a—Tracheal lobe
   b—The so-called lower accessory lobe
   c—Ectopic lung.

The rarity of agenesis of the lung is attested to by the infrequency with which it has been reported in the literature. As of January 1, 1954, there were less than 150 cases reported in medical journals. In a review of 100 cases (96 from the literature and four reported in this paper) there were 40 males and 38 females. In 22 the sex is not mentioned. The right lung was involved in 48 and the left in 52. The age distribution in 86, in which it was mentioned, was as follows: premature, seven; birth to one week, six; one week to one month, eight; one month to three months, three; three months to six months, nine; six months to one year, six; one year to two years, 10; two to five years, eight; five to 15 years, six; 15 to 25 years, eight; 25 to 35 years, three; 35 to 50 years, six; 50 to 65 years, four, and 65 to 72 years, two.

Symptoms in patients with congenital pulmonary agenesis are variable, from complete absence to those of marked cardiorespiratory embarrassment. When symptoms do occur, they are not necessarily due entirely to the agenesis, but may also be due to associated malformations and complications. The most frequent symptoms encountered in patients with

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clinical manifestations of pulmonary agenesis are dyspnea and tachycardia, occasionally accompanied by stridor and cyanosis. The commonest thoracic physical findings are symmetry of the chest and some respiratory lag of the affected side. The involved side usually is not smaller than the uninvolved side. Such findings in the young subject with progressive respiratory distress, should make one highly suspicious of agenesis of the lung. This is particularly true when it is accompanied by displacement of the heart, trachea, and mediastinum to the affected side and there is elevation of the diaphragm on that side. The degree of displacement of these structures varies with the degree of pulmonary agenesis. Roentgenographic studies of the thorax confirm physical findings. Absence of asymmetry of the chest and displacement of structures as mentioned in the foregoing are cardinal findings in pulmonary agenesis. Bronchoscopic and bronchographic variants are many. In general, however, they disclose rudimentary development of the bronchial tree on the affected side, and distortion and displacement of the bronchial tree on the uninvolved side.

Case 1: A five pound, eight ounce newborn female infant, delivered spontaneously at full term, began to show signs of respiratory distress soon after birth. Accessory muscles of respiration were very active. Breath sounds were harsh and accompanied by crackling rales over the right hemithorax. Heart sounds were distant and heard best in the right hemithorax. A roentgenogram of the chest revealed the heart and mediastinum in the right hemithorax and a diffuse nonaerated opacity over the greater part of the upper right lung field, Fig. 1.

Bronchoscopy revealed the trachea and both main bronchi obscured by frothy clear mucus. Except for some displacement of the left main bronchus to the right, it was otherwise not remarkable. The right main bronchus, on the other hand, appeared to be a direct downward continuation of the trachea. The orifices of the right upper and middle lobes could not be identified. The child improved after bronchoscopy, but died in respiratory distress 48 hours after birth.

At necropsy, the left lung was essentially normal, although somewhat distended. The right lung was constituted of but one lobe having a single bronchus which took the course of a normal right lower lobe bronchus, Fig. 2. The intrapulmonic subdivisions of the bronchus corresponded to that of a normal right lower lobe bronchus.

**Figure 1**: Opacity of the upper two-thirds of the right hemithorax with retraction of the heart and mediastinum into that area as is characteristically found in agenesis of the (right) lung. Same case as Fig. 2.—**Figure 2**: Anterior view of the intrathoracic contents as seen at necropsy in agenesis of the right lung. The heart is completely displaced to the right of the midline. The right lung is comprised of only the lower lobe. Same case as Fig. 1.
and did not present any evidence of an intrapulmonic subdivision of the right main bronchus.

The esophagus was constituted of two separate segments. The upper portion formed a continuation of the pharynx in the normal manner, but ended blindly at about 1 cm. from its origin. The lower segment originated in the region of the bifurcation of the trachea and ended by opening into the stomach below. The kidneys were fused to form a solitary horseshoe kidney.

Case 2: A seven year old child entered the hospital with a diagnosis of situs inversus. According to the mother, the heart was known to be on the right side for some time.

Examination revealed a well nourished, well developed, cooperative male child. The trachea was deviated to the right of the midline. There was dullness to percussion over the greater part of the right anterior part of the thorax. In the same area the breath sounds were distant and bronchial. The point of maximum cardiac pulsation was 5 cm. to the right of the midsternal line in the third and fourth intercostal spaces. Roentgenographic examination of the chest showed the heart retracted into the right hemithorax, the trachea deviated to the right, the left lung was emphysematous, and the right hemithorax was somewhat smaller than the left. Bronchoscopy showed nothing remarkable in the left bronchial tree. However, on the right side there was absence of the upper and middle lobe bronchi. The bronchus to the lower lobe was wider than normal, but otherwise was not remarkable.

Case 3: A 19 year old man entered the hospital for bronchoscopy because he had a "shadow in the chest". This condition was discovered during a routine x-ray film examination of the chest. The only significant history relative to the patient's thorax was that he would cough and had mucopurulent expectoration after exercise.

Examination revealed the following positive findings: (1) increased dullness to percussion over the right side of the chest both anteriorly and posteriorly, (2) increased tactile and vocal fremitus over the upper right half of the chest posteriorly, (3) bronchial breathing and hyper-resonance to percussion over the left side of the thorax, and (4) the point of maximum cardiac impulse was in the right anterior fourth intercostal space. The admission roentgenogram of the chest disclosed narrowing of the intercostal spaces on that side. The heart and trachea were completely within the right hemithorax.

In view of the history and x-ray findings it was thought that he had a nonopaque foreign body in the bronchus and that he should be bronchoscoped. Accordingly, this was performed but was unsuccessful because of the copious secretions in the right main bronchus which could not be removed satisfactorily by repeated aspirations via the bronchoscope. Bronchoscopy reinstituted after several days of postural drainage revealed a moderate amount of mucopurulent secretion in the left main bronchus. The subdivisions of the left bronchial tree were otherwise negative. The right main bronchus also contained a moderate amount of mucopurulent secretion. When these secretions were removed it was noted that the orifice to the lower lobe was markedly stenosed by mucosal edema. The upper and middle lobe orifices could not be visualized.

Tomography after instillation of lipiodol into the bronchial tree on the right side disclosed marked angulation of the right main bronchus posteriorly and substantiated the diagnosis of absence of the bronchi to the upper and middle lobes on the right side, Fig. 3. In view of the fact that the roentgenograms did show lung tissue in the upper part of the right hemithorax, although unaerated and possible nonfunctioning, it was deemed advisable to explore the thorax and remove the pathological tissue, if found. At the time of the operation it was noted that the right upper and middle lobes were absent and that the superior segment of the lower lobe was emphysematous, the basilar segment was indurated and contained many cystic areas. Concurrently with the absence of the upper and middle lobes the arteries and veins to those lobes were also absent.

Case 4: A 35 year old man entered the hospital in September 1952 in a moribund condition as a result of a head injury sustained in an automobile accident. He died soon after admission. Necropsy, Fig. 4, revealed absence of the right lung. At the site of the supposed origin of the right main bronchus, there was a heaping up of bronchial tissues.

CONCLUSIONS

Although pulmonary agenesis is an uncommon condition, it warrants consideration in all patients whose clinical course is not readily explainable on some other more satisfactory basis. This is particularly true in younger subjects. The frequency with which one encounters other mal-
formations in patients with pulmonary agenesis has led Ferguson and Neuhauser to explain the genesis of this condition on the basis of a defect in the germ plasm. Wexels observed that nearly 50 per cent of the patients with pulmonary agenesis have other defects. He lists these anomalies as—patent ductus arteriosus, patent foramen ovale, atresia ani, atresia of the esophagus, short bowel, atresia urogenitalis, exencephaly, horseshoe kidney, ventricular defect, hypoplasia of the face, hare lip and cleft palate, absence of the left diaphragm, absent left hand, absence of right radius, bifid uvula, spina bifida, wedge-shaped vertebrae, hemivertebra, deformed ribs, accessory thumb, closed foramen epiploica, rudimentary atlas, deformed external ear, congenital ptosis of both eyelids, small left scapula, and pulmonary veins entering the azygos vein.

Pulmonary agenesis is not incompatible with life. Twenty-three patients (27 per cent) of the present series were known to have lived to be between 15 and 72 years. In reviewing the literature regarding age and agenesis, we were impressed with the fact that those who were known to have pulmonary agenesis in early life survived it only because the associated anomalies and complications were not incapacitating. Agenesis in the patient over 15 years of age is, in all likelihood, unassociated with other malformations which are of clinical significance. On the other hand, pulmonary agenesis in early life, which is clinically manifest, is often associated with other malformations which make the condition incompatible with life.

All patients who are suspected of having pulmonary agenesis should be investigated thoroughly before being subjected to any thoracic surgical procedure. A relatively minor procedure in such a patient may terminate fatally because of the lack of appreciation of the significance of the other anomalies.

**FIGURE 3**

*Figure 3:* Roentgenogram of the chest in a patient with pulmonary agenesis of the right side.—**FIGURE 4**

*Figure 4:* Anterior view of the intrathoracic contents in a patient with agenesis of the right lung.
CONCLUSIONES

Aunque la agenesia pulmonar es un padecimiento poco común, amerita ser considerado en todos aquellos pacientes cuyo curso clínico no es fácilmente explicable sobre otras bases más satisfactorias. Esto es particularmente cierto en los sujetos más jóvenes. La frecuencia con que uno encuentra otras malformaciones en pacientes con agenesia pulmonar, ha llevado a Ferguson y Neuhauser a explicar la génesis de este padecimiento sobre las bases de un defecto en el plasma germinal. Wexels observó que muy cerca del 50% de los pacientes con agenesia pulmonar tienen otros defectos. Cita las siguientes anormalidades: persistencia del conducto arterioso, persistencia del agujero oval, atresia anal, atresia esofágica, agenesia intestinal, atresia urogenital, anencefalia, riñón en herradura, defectos ventriculares, hipoplasia de la cara, labio leporino y paladar hendido, ausencia del diafragma izquierdo, ausencia de mano izquierda, ausencia de radio derecho, úvula bifida, espina bifida, vértebras en cuña, hemivértebras, costillas deformadas, pulgar supernumerario, ausencia de hiato de Winslow, atlas rudimentario, deformación del oído externo, ptosis palpebral congénita, escápula izquierda pequeña y venas pulmonares que desembocan a la vena acigos.

La agenesia pulmonar no es incompatible con la vida. Veintitrés pacientes (27%) de las series que ahora se presentan se sabe que vivieron hasta llegar de 15 a 72 años. Revisando la literatura respecto a edad y agenesia, nos ha impresionado el hecho de que aquellos en quienes se sabía presentaban agenesia pulmonar en los primeros años de su vida, sobrevivieron a ésta sóloamente porque las anomalías asociadas y complicaciones no fueron incapacitantes. La agenesia en un paciente mayor de 15 años de edad con toda posibilidad no está asociada con otras malformaciones que sean de significado clínico. Por otro lado, la agenesia pulmonar a temprana edad que es manifiesta clínicamente, está a menudo asociada con otras malformaciones que hacen el padecimiento incompatible con la vida. Todos los pacientes sospechosos de tener agenesia pulmonar deben ser investigados minuciosamente antes de ser sometidos a cualquier procedimiento quirúrgico de tórax. Un procedimiento quirúrgico relativamente pequeño en tales pacientes puede terminar fatalmente, debido a la falta de apreciación de la magnitud de las otras anomalías.

RESUME

Bien que l'agénésie pulmonaire soit un état peu habituel, elle demande à être prise en considération chez tous les malades dont les manifestations cliniques ne peuvent pratiquement pas s'expliquer par une autre cause plus satisfaisante. Ceci est spécialement vrai chez les sujets jeunes. La fréquence avec laquelle on rencontre d'autres malformations chez les malades atteints d'agénésie pulmonaire a conduit Ferguson et Neuhauser à l'expliquer par un défaut du plasma germinatif. Wexels observa que près de 50% des malades atteints d'agénésie pulmonaire présentaient d'autres malformations. Il les dénombra: persistence du canal artériel, persistance du trou de Botal, atrésie anale, atrésie oesophagienne, brièveté intestinale,
l'agenésie pulmonaire n'est pas incompatible avec la vie. 23 malades (27%) du groupe étudié vécurent de 25 à 72 ans. En faisant la revue de la littérature en ce qui concerne l'âge et l'agenésie, les auteurs furent frappés du fait que ceux qui étaient connus comme ayant une agénésie pulmonaire depuis l'enfance, survécurent parce que les anomalies et complications associées n'entraînaient pas une impossibilité d'existence. L'agenésie chez le malade de plus de 15 ans n'est pas associée, en général, à d'autres malformations cliniquement notables. Pendant le premier âge, l'agenésie pulmonaire cliniquement évidente, est souvent associée à d'autres malformations qui rendent l'état incompatible avec la vie.

Tous les malades suspects d'être atteints d'agenésie pulmonaire devraient être examinés complètement avant que soit tentée toute intervention chirurgicale thoracique. Une opération relativement peu importante peut avoir, chez un tel malade, une conséquence fatale, parce qu'on n'a pas apprécié la valeur des autres anomalies.

BIBLIOGRAPHIES


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