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Short Rib-Polydactyly Syndrome (Majewski Type): Report of a Case with Autopsy Findings

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ABSTRACT

An autopsy case of short rib-polydactyly syndrome, type II (Majewski type) complicated by anisosplenia is reported, and some discussion has been made from the feto-pathological viewpoint on the charactaristic findings and concomitant malformations of this syndrome with regard to a total of 23 cases, including our case and 22 cases reported in literature.

Short rib-polydactyly syndrome, type II was first proposed by Majewski et al[8)] in 1971 as a type of lethal short-limbed dwarfism presenting the finding of complication by cleft lip and palate, narrow thorax, short extremities, polydactyly, multiple visceral abnormalities, etc. The cases reported subsequently are only 2 by Motegi et al[4,9-11)] and 1 each by Spranger et al[18)], Kodama et al[5,6)] and Goto et al[2)]. As evident from this, this anomaly is rare among lethal short-limbed dwarfisms. The type complicated by cardiovascular anomalies is not described at all in reports that have been made on this type in Japan. The authors have autopsied a case in which cystic pancreas fibrosis, cardiovascular anomalies and anisosplenia[7)] were associated with this syndrome, and we wish to study this type of malformation together with the cases mentioned above.

REPORT OF A CASE

Familial history: Father is 29 years old and mother, 24 years old. Both are healthy, and their past histories are unremarkable. No blood relationship including near relationship nor heredopathia have been found. Both paternal and maternal grand parents have no experience of atomic bomb exposure. Mother's first child (at age 21) terminated in an artificial abortion, and the subsequent pregnancy is the present pregnancy.

Course of labor and delivery: The last menstruation lasted for 5 days from 26 December 1982. There was no history of drug-taking or irradiation during pregnancy. The course of the pregnancy was normal. Early rupture of membrane occurred on 11 August 1983, and a baby girl was born in spontaneous delivery in the 9th month of pregnancy (33rd week). Because the baby was a still-birth with placenta accreta, the placenta was extracted 2 days after childbirth.

AUTOPSY FINDINGS

Measurement: The weight of the infant was 2,300g; crown-heel length, 36cm; crown-rump length, 27cm; head circumference, 30cm; chest circumference, 30cm; abdomen circumference, 35cm; upper extremities, both right and left; 10.5cm; lower extremities, both right and left; 12.0cm.

External views: Hydrops anasarca and short extremities were remarkable. Especially, the

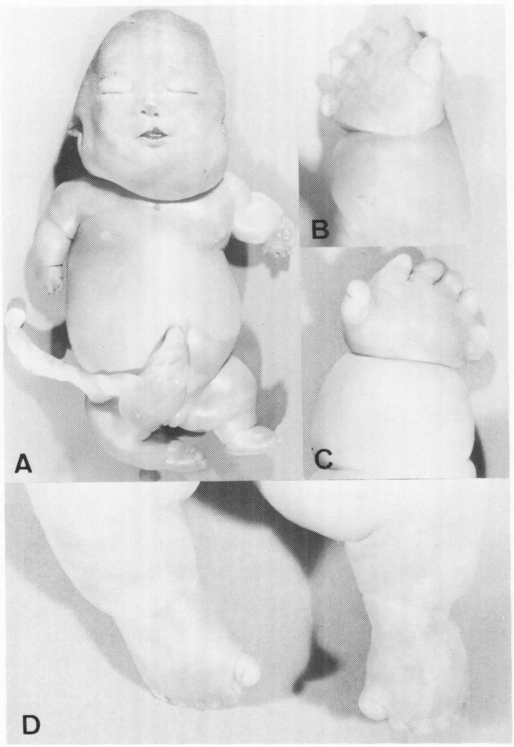

Fig. 1. Postmortem photographs of the specimen. A: Anterior view showing the universal hydrops. B and C: Right and left hands showing postaxial polydactyly and syndactyly (lt.) D and E: Lower extremities showing preaxial polydactyly of the toes.

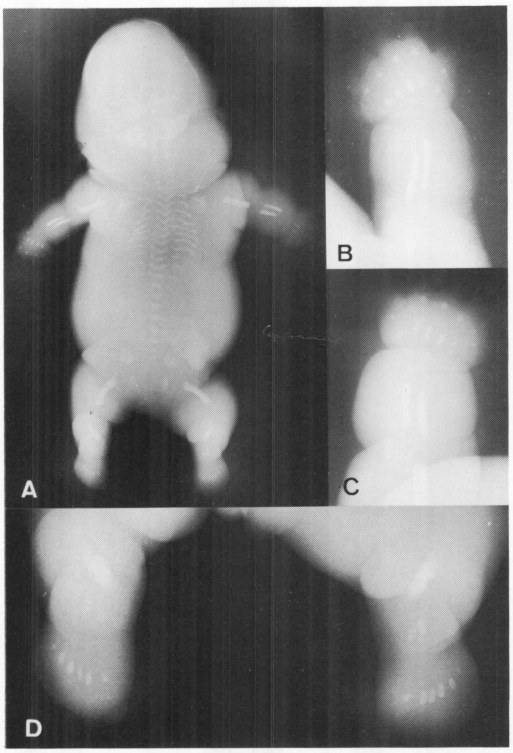

Fig. 2. Radiographs of the specimen. A, B, C and D: Corresponds to that of A, B, C and D in Fig. 1, respectively (see text).

edema extending from the left mandibular region to the neck presented a tumor-like appearance. Sparse hair, short neck, saddle nose, incomplete upper cleft lip (left), and low set ears were noted. The abdomen was protuberant and the umbilical cord presented a cystic appearance (Fig. 1A). Both hands had 6 digits each (Figs. 1B and C, Figs. 2B and C), and the 6th finger of the left hand presented skin-like syndactyly. This anomaly was considerd to be postaxial polydactyly and brachydactyly. Both feet presented preaxial polydacty with 6 digits each, and both showed incomplete syndactyly of the 3rd-5th toes of the feet with marked brachydactyly (Fig. 1D, Fig. 2D).

Internal organs: Excepting intestinal compression of the right abdominal cavity, the situs of organs was normal (solitus). Maxillary incisor tooth formation (2 teeth), hypoplasia of larynx, marked narrow thorax due to short ribs (Fig. 2A, Figs. 3A and B), excessive hypoplasia of the lungs, incomplete lobation of the left lung, hepatomegaly complicated by abnormal lobation and partial fibrosis (Fig. 3A, arrow), hypoplasia of the gall bladder, cystic pancreas fibrosis with a large cyst (4 × 4cm) in the pancreas tail (Fig. 3B, Fig. 5), renal hypoplasia with multiple kidney cysts, accessory spleens (23) of miliary size to soy bean size and incisura formation (Fig. 3B) of the spleen, hypoplasia of the ovary, and serous ascites (Ca 20cc) were found. The heart showed complete atrial septal defect (incomplete persistent atrio-ventricular canal), hypoplastic aorta with bicuspid aortic valves, elongation and narrow outflow tract of the left ventricle, and persistent left superior vena cava (Figs. 4A and B).

The brain showed slight hypoplasia.

RADIOGRAPHIC FINDINGS

The longs bones were all remarkably short, and some epiphyseal line of the long bones were observed to be slightly irregular. The tibia, in particular, was strikingly short and elliptic. On X-ray, the ossificated ribs were exceedingly short and horizontal, and the chondro-osseous border of its anterior ends, that is the rib epiphysis, was observed to be enlarged like a club. Each of the 6 pairs of phalanges showed hypoplasia, and the scapula and the pelvic bone also showed slight hypoplasia. The skull and the vertebrae were unremarkable. No osscification center was found on the femoral head (Figs. 2A, B, C and D).

HISTOLOGICAL FINGINGS

Pancreas: A remarkable fibrous proliferation of the interstitial connective tissues and dilatation of the ductus were found. Hardly any islets of Langerhans were observed. There was dilatation of the acinous lumen, but it was not pronounced (Fig. 7). The internal wall of the part that had formed a large pancreas cyst was covered with a layer of pavement cells, but the wall was made up of connective tissues and acini were found embedded in them (Fig. 6).

Kidney: Mild proliferation, hyperemia and hematopoiesis were found in the interstitial connective tissues. Urinary tubules of irregular sizes were dilatated and had formed cysts. Degeneration and sclerosis of glomeruli were seen in some of them (Fig. 8).

Liver: There was marked fibrosis and proliferation of dilatated bile ducts, and also abundant hematopoiesis, in the periphery of the portal vein. Numerous premature eosinophil cells were seen in some parts. The hepatic cell cords were irregularly arranged, the sinusoid was dilatated, and hematopoiesis of erythroblasts was active (Fig. 9).

Thyroid: The follicles were of various sizes and irregular shape and were covered with a layer of cuboidal cells, but there was no colloid formation (Fig. 10).

Lung: Mild angiectasis was seen in the interstitial connective tissues, but there was no dilatation of the bronchus or the alveolus (Fig. 11).

Sternum: The bone surface was uneven and the columnar cartilages were irregularly arranged in some parts (Fig. 12).

Others: The pituitary gland, thymus, adrenals, ovary, myocardium and spleen showed no remarkable changes except hyperemia.

DISCUSSION

The frequency of lethal short-limbed dwarfism in newborns is said to be 1 in 19000[1]. Thirty cases of lethal short-limbed dwarfism were found in 10,834 autopsy cases of fetuses and newborns. Among them, one was a case of short rib -polydactyly syndrome, Majewski type (approx. 1 in 10000)(Hayashi and Okamoto, 1984)[3]. Although it is classified in the category of osteochondrodysplasia in the Nomenclature of Constitutional Disease of Bone (1978)[17], short rib-polydactyly syndrome is, as mentioned above, a very rare disease. At present, this anomaly is classified into two types[23], the Saldino-Noonan type and the Majewski type, but a report has been made recently (Naumoff, 1977)[12] which suggests, from the fact that the type of anomaly of the cervical vertebrae and pelvic bone differs from the above two types, that there is a third type. However, the International Nomenclature classifies the syndrome into only two types, type I (Saldino-Noonan type) and type II (Majewski type)[22].

As already mentioned, short rib-polydactyly syndrome, type II, was reported as a congenital malformation syndrome by Majewski et al (1971)[8] from their study of 17 cases including 4 cases experienced by themselves and 13 cases reported in literature. Subsequent to the report of Majewski et al only 5 cases have been reported, 1 by Spranger et al (1974)[18], 2 by Motegi et al (1977[9], 1979)[10,11] and 1 each by Kodama et al (1978)[5] and Goto et al (1983)[2]. Hence, only 23 cases of the Majewski type have been reported thus far including the authors' case.

The characteristic findings of this type with respect to the 5 cases reported subsequent to the report of Majewski et al and the authors' case are shown in Tables 1 and 2.

With 3 cases being male and 3 cases female, no difference by sex was noticed. As regards the

Table 1. Summary of previously reported and present cases of short rib-polydactyly syndrome (Majewski type)

	Spranger et al[18]	Motegi et al[9,11]		Kodama et al[5]	Goto et al[2]	authors
		Case 1	Case 2			
Sex	F	M	M	M	F	F
Gestation (weeks)	unknown	37	37	37	35	33
Body weight (g)	3500	3150	2410	3150	1780	2300
Crown-Heel length (cm)	38.1	45	42	45	27	36
Neonatal mortality	unknown	11 hr	25 hr	still birth	soon	soon
Hydrops	+	+	+	+	+	+
Cranio-oro-facial anomalies: cleft lip	+	+	+			+
Cranio-oro-facial anomalies: cleft palate	+				+	
Cranio-oro-facial anomalies: larynx hypo.		+	+			+
Cranio-oro-facial anomalies: micrognathia	+			+		
Cranio-oro-facial anomalies: low set ears	+	+	+	+	+	+
Cranio-oro-facial anomalies: facial fissure				+		
Protuberant abdomen	+	+	+	+	+	+
Short extremities	+	+	+	+	+	+
Poly-syn-brachydactyly	+	+	+	+	+	+
Hydramnion	+	+	+			

Table 2. Summary previously reported and present cases of short rib-polydactyly syndrome (Majewski type)

	Spranger et al[18]	Motegi et al[9,11]		Kodama et al[5]	Goto et al[2]	authors
		Case 1	Case 2			
Narrow thorax	+	+	+	+	+	+
C.N.S. anomaly						+
Cardiovascular anomaly						+
Hypoplasia, lungs	+	+	+	+	+	+
Digestive anom.: mesenterium comm.				+	+	
Digestive anom.: hepatomegaly	+			+	+	+
Digestive anom.: pancreas fibrosis					+	+
Urino-genital anom.: renal hypoplasia					+	
Urino-genital anom.: cystic kidney	+	+	+	+		+
Urino-genital anom.: int. genital anom.		+		+	+	+
Urino-genital anom.: ext. genital anom.		+	+			
Others						anisosplenia

time of death, the cases were either still-birth or death within two days postnatally. Hydramnion is reported in 3 cases[11,18].

No report made any mention concerning the placenta, but in the authors' case, it weighed 390g and was rather small compared with that of cases of the same fetal age, placenta accreta was present, and protracted separation was observed.

As Table 1 shows, hydrops anasarca, narrow thorax, brachy-syn-polydactyly, and various types of cranio-oro-facial anomalies were observed in every one of the six cases.

Among these, the cranio-oro-facial anomaly that was observed in every case was low set ears, and half of them were complicated by auricular hypoplasia or abnormality. Association of cleft lip with cleft palate, upper middle or upper left cleft lip, saddle nose, and hypoplasia of pharynx and larynx were observed in four cases. Besides, one case each of cleft palate and ocular hypertelorism associated with facial cleft (macrostomia) and micrognathia and teeth abnormality were observed. Rarely, case of no complication by hydrops anasarca is reported[8].

The short extremity was symmetrical in every case, and it was remarkable in the lower extremities. Polydactyly consisted of 6-8 digits, and both preaxial and postaxial types were found. Besides, some were associated with brachy -syndactyly. The largest number of digits in polydactyly was 10 digits[8]. Remarkable short rib and long bone and short elliptical tibia were observed in the ossal system of all cases. Histologically, these cases presented irregular arrangement of chondrocytes in the columnar cartilage and decreased proliferation of bone cells in the epiphyseal cartilage region. A case has also been reported in which there was a residuum of cartilage in the bone trabecula[2].

Radiographic findings: 1) In all cases, the rib was short and horizontal with enlargement of the anterior end. One case was found in which short rib was not presented on radiography (Majewski et al, case 4)[8]. Neither was narrow thorax described for this case. However, narrow thorax can be surmised to have been present from the postmortem examination record which described the lung as being exceedingly hypoplastic with no dilatation. Some cases also showed cap-like or club-like changes of the rib end[2,5,11]. 2) The long bones were observed to be short, especially the tibia, which was exceedingly short and elliptic, but the finding of irregular epiphyseal line of the long bone was rare. The finding of ossification of the proximal epiphyses of the humerus and the femur in some cases is also reported[11,18]. 3) As regards the phalanges, some cases showed the 4th and 5th metacarpalia of the right hand to be shaped like the letter Y, and the presence of osseous defect of the digital metacarpalia or the 7th finger of the left hand[2,4]. 4) The skull and the vertebrae were normal in all the cases. 5) Dysplasia of the pelvic bone has been reported in some of the cases (Majewski et al, cases 1, 2 and 4)[8].

An abnormality of the urino-genital system of some kind was found in every one of the 6 cases. With the exception of one case[11] of doubtful description (hermaphroditism ?), retentio testes was found in 2 of the 3 male cases[6,11]. In the female cases, a case each of uterus bicornis[2] and hyperplasia of both overies were fond. Generally, these genital anomalies are frequently associated with the Majewski type, and there are reports of cases in females, besides those described above, in which incomplete vagina septa (Majewski et al, case 3)[8] and vagina septa (Majewski et al, case 1)[8] were associated with uterus bicornis.

Multiple kidney cysts were found in 4[5,11,18] of the 6 cases, and one of the cases was described as polycystic kidney[5]. Besides, the cases of renal hypoplasia with or without hydroureter and cartilage formation were also found[2].

According to the description of Majewski et al, cardiovascular anomalies were observed as complications in 8 of 13 cases (61.5%)[8]. However, in the present study, the authors' case was the only one among the 6 cases that was complicated by a cardiovascular anomaly. Five of these 6 cases are cases reported in Japan, which gives us to conjecture the presence of a racial difference. This should be ascertained by collecting a larger number of cases.

As for anomalies of the spleen, the authors' case was the only one that presented anisosplenia (Landing, 1971)[7], an anomaly in which a large number of accessory spleens are present, but a case which showed splenomegaly has been reported[2]. Since it is well known that there is a relationship between splenic anomalies and

Table 3. The ratio of associated malformation to short rib-polydactyly syndrome (Majewski type)

	Majewski et al[8]	authors
Sex ratio (female/male)	5/9	3/3
Perinatal death	17/17	6/6
Hydrops	14/16	6/6
Short extremities	17/17	6/6
Polydactyly	17/17	6/6
preaxial	12/14	5/6
axial	13/14	2/6
postaxial	13/14	6/6
7-10 fingers/toes	14/17	3/6
Syndactyly	15/16	5/5
Brachydactyly	17/17	6/6
Cranio-oro-facial anom. cleft lip	8/16	4/6
Cranio-oro-facial anom. cleft palate	5/16	2/6
Cranio-oro-facial anom. low set ears	5/6	6/6
Cardiovascular anomaly	8/13	1/6
Liver anomaly	8/13	4/4
Urino-genital anom. renal hypoplasia	2/13	1/1
Urino-genital anom. cystic kidney	3/13	5/6
Urino-genital anom. genital anomaly	11/14	5/6
Hydramnion	1/1	3/6

cardiovascular anomalies[3,13,14], the authors' case presumably has a relationship to the formation of anisosplenia.

Hypoplasia of the lungs was observed in all of the cases. This possibly is ralated to the development of narrow thorax. Besides, one case each of aspiration pneumonia (Majewski et al, case 4)[8] and hyaline membrane disease[11] have been reported.

As regards anomalies of the digestive system, hepatomegaly was observed in all the cases excepting two[11] in which this was not described. The hepatomegaly was especially remarkable in the left lobe, extending as far as the left abdominal wall in some cases, and this was observed to be complicated by abnormal lobation in some cases. Further, a picture of retained extramedullary hematopoiesis was observed in some cases (Majewski et al, cases 1, 3 and 4)[8]. Histologically, the authors' case showed fibrosis of the peripheral connective tissues of the portal venous system and proliferation of the bile duct. This finding presumably is related to cystic pancreas fibrosis which will be discribed later. Two cases complicated by mild proliferation of bile ducts in the portal vein have been reported (Majewski et al, cases 1 and 3)[8], but these cases showed no complication by cystic pancreas fibrosis as observed in the authors' case.

One case complicated by hepatomegaly and absence of the gall bladder has been reported (Majewski et al, case 2)[8]. That the liver is frequently reported as presenting hematopoiesis on histological examination suggests the presence of retardation in this malformation compared with fetuses of the same fetal age.

The pancreas of the authors' case showed advanced fibrosis with large cyst. As this cystic pancreas fibrosis is known to be an autosomal recessive inheritance[15], its possible relationship to the cause of short rib-polydactyly syndrome is suggested.

In addition, 3 cases of mesenterium communis and 1 case each of intestinal malrotation and short intestine were found.

As regards the central nervous system,

cerebral hypoplasia was found only in the authors' case among the 6 cases. However, an anomaly in which cerebellar hypoplasia was associated with flat gyrus and absence of the bulbus olfactorius (Majewski et al, case 1)[8] has already been reported.

Others: A biochemical examination of this malformation was made using umbilical blood, and the values of Ca, P, and Al-p which are related to bone formation were each reported to be within normal range[5]. Blood gas analysis of case 2 of Motegi et al, which showed an Apgar score 3 at birth and lived for 25 hours after birth, presented respiratory acidosis with decreased O_2 and accumulated CO_2 due to marked ventilatory impairment, showing, with administration of O_2 100%, the values of pH 7.043, PO_2 39.9 mmHg, PCO_2 68.7mmHg, BE −14.2 mEq/liter, and O_2 sat 51.3%.

Otherwise, except for a slight decrease of immunoglobulin G, mineral, urea N, ECG and cytogenetic study were all reported as being normal[11]. Goto et al conducted acid mucopolysaccharide analysis using 6 kinds of bones or cartilages and, confirming that the bone has a high uronic acid content and contains a large quantity of chondroitin sulfuric acid C, assumed that the distribution of acid mucopolysccharide approximated that of the chondral type and that such abnormality of mucopolysaccharide metabolism disturbed the transformation of cartilage into bone in the ossification process[2].

The cause, which was reported to be unknown[19,24], has come to be attributed to autosomal recessive inheritance[23] following the report of Motegi et al[4,11] on siblings. Autosomal recessive inheritance is suspected in the authors' case also as it was complicated by cystic pancreas fibrosis. On the other hand, the chromosomes of two cases had been studied[5,11] and no chromosome aberration was found in either of them.

Cause of death: As mentioned earlier, respiratory insufficiency (pulmonary functional insufficiency) arising from marked hypoplasia and atelectasis of the lungs can be considered to be the foremost cause. This finding was common to all of the cases.

Differential diagnosis: Short rib-polydactyly syndrome, type II (Majewski type), which presents such a syndrome as described above,[19,21,23-25] needs to be differentiated from type I (Saldino-Noonan type)[16]. Concerning this, detailed reviews have been made by Tsuruta[23], Tsuruta and Sugiura[25], Sugiura and Tsuruta[19-21], and Tsuruta et al[24].

Narrow thorax, short extremities, and multiple visceral abnormalities are found alike in both types. On radiography, short rib in horizontal position and enlargement of its anterior end and short long bones are found common to both types.

As morphological differences between the two types, anal atresia is frequently seen, and polydactyly only postaxially, in the Saldino-Noonan type, but cleft lip and palate are frequent and polydactyly occurs preaxially, postaxially or axially in the Majewski type. On radiography, the Saldino-Noonan type shows spinous unevenness (candle flame)[18] of long bone epiphyses, ossification of the proximal epiphyses of long bones, and flattening of vertebrae due to poor ossification, but the Majewski type shows a relatively smooth long bone epiphyseal line, with little ossification of the proximal epiphyses. Further, findings such as an especially short and elliptical tibia and nearly normal vertebrae are frequently observed.

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Fig. 3A. Macrophotograph of the thoracic and abdominal organs showing narrowed chest, enlargement of liver (L) with partial portal cirrhosis (arrow) and large pancreas cyst (P). H: heart, IT: intestine.

Fig. 3B. Macrophotograph of the abdominal organs showing cystic pancreas fibrosis (P) and anisosplenia (SP). ST: stomach, O: ovarium, UT: uterus, UB: urinary bladder, IT: intestine.

Fig. 4A. External configuration of the heart showing relationships between the aorta (AO) and pulmonary trunk (PT). AT: atrium, RV: right ventricle.

Fig. 4B. Left ventricular view showing elongated and narrowed left ventricular outflow tract (OT) in the atrio -ventricular septal defect (arrow). AO: aorta, LV: left ventricle.

Fig. 5. External configuration of the pancreas showing large pancreas cyst (C).

Fig. 6. Histological finding of the wall of pancreas cyst. Increased interstitial connective tissues and scattered acini are seen.

Fig. 7. Pancreas. Interstitial connective tissue is conspicuously increased. Lumens of acini are somewhat dilated and no acidophilic hyaline-like materials were found.

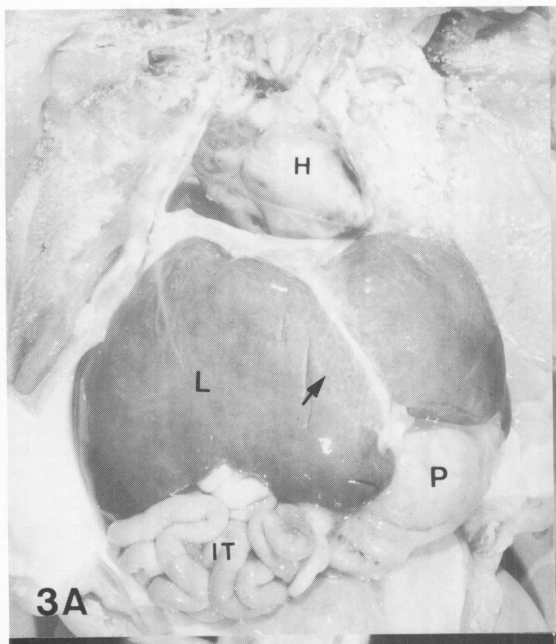
Fig. 8. Kidney showing pronounced dilatation of urinary tubule. Mild degeneration of glomeruli and fibrosis of Bowman's capsules are noted (arrow).

Fig. 9. Liver showing extreme portal fibrosis with dilated and proliferated bile ducts (arrow).

Fig. 10. Thyroid. Acini are irregular in size and no colloid is present.

Fig. 11. Lung showing reduced differentiation of alveolar ducts and alveoli.

Fig. 12. Sternum showing irregular bone surface and columnar cartilage.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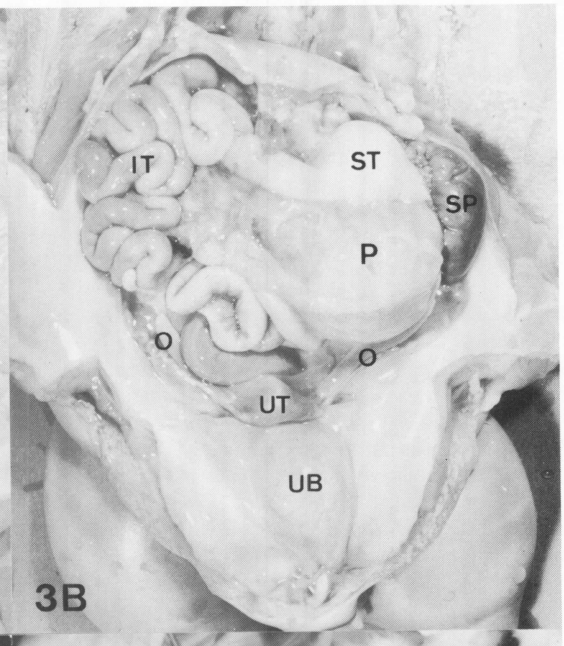
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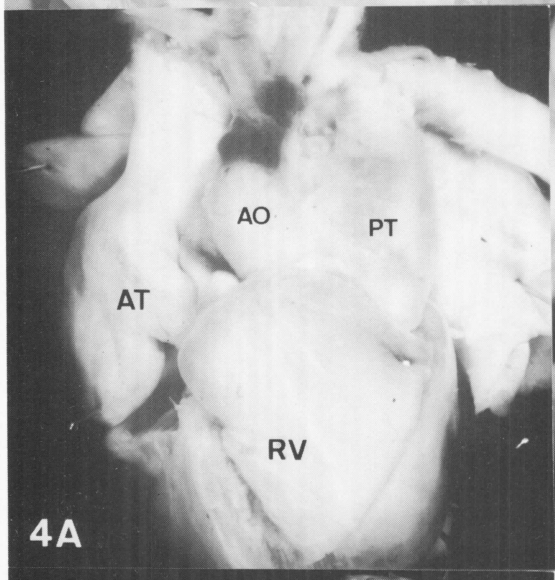
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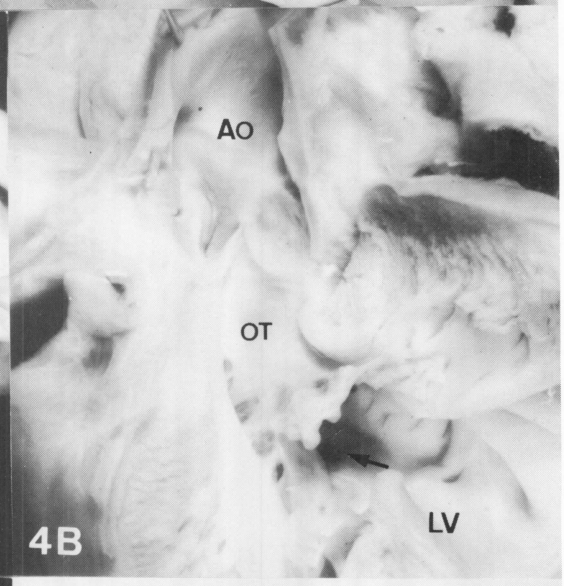
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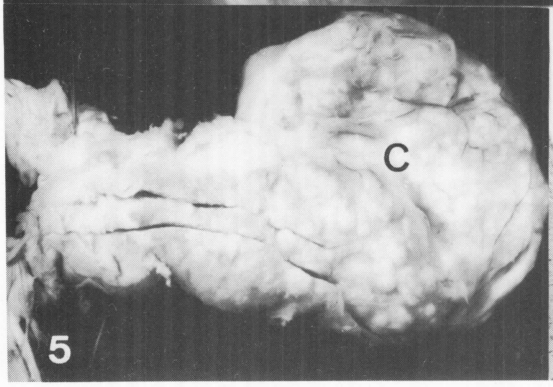
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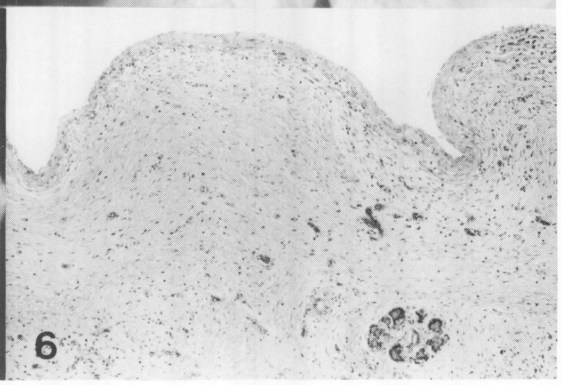
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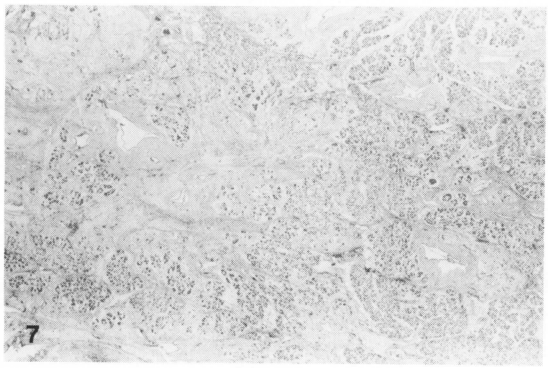
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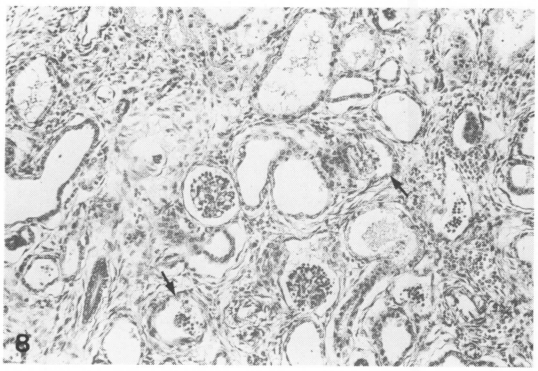
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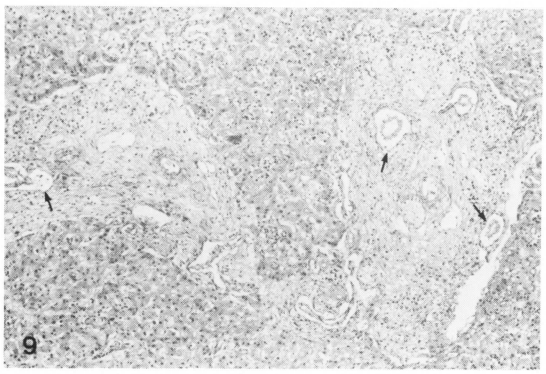
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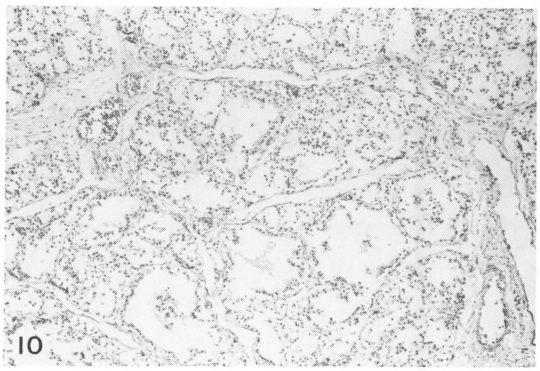
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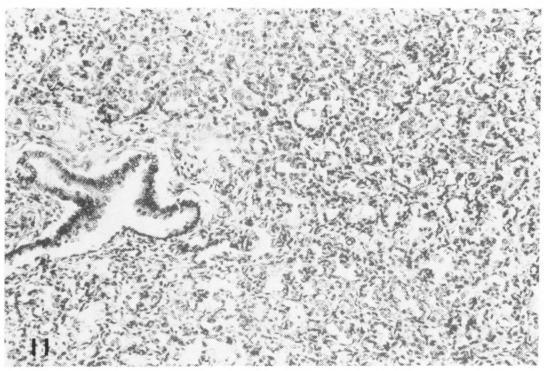
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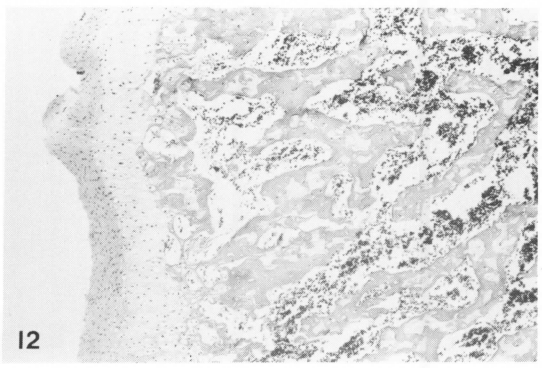
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