MALFORMATION OF EARS AS SIGN OF MALFORMATION OF GENITO-URINARY TRACT

BY

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During the past seven years a number of patients have been encountered with deformities of the external ear associated with congenital abnormalities of the genito-urinary tract. In all of them it was the presence of the deformed ear that drew attention to the possibility of underlying malformation of the genito-urinary tract as an aetiological factor in the clinical picture.

Three newborn infants were seen with the typical facial features of renal agenesis so lucidly described by Potter (1946). She describes the facies thus: "The most striking feature consists of an increased width between the eyes, and the presence of an unusually prominent fold arising at the inner canthus of each eye. The fold sweeps downwards and laterally to form a wide semicircle under the inferior medial aspect of each orbital space. Other changes which, when combined with the appearance of the eyes, give the face of the infant a resemblance to that of a person of very advanced age—an extreme premature senility as it were—are a flattening and slight broadening of the nose, an unusually receding chin, and large low-set ears which have proportionately little cartilage" (see Fig. 4). Although two of the three infants seen had ears which conformed to Potter's description, the third had small folded ears, set low, rotated backwards, and a bulbous-edged helix which was sliced or "squared off" across its upper edge (see Fig. 8, lower inset picture). A fourth infant whose facies conformed completely to Potter's description survived.

It has become apparent that malformed ears, particularly if asymmetrical, are quite commonly associated with congenital malformations of the genito-urinary tract often compatible with life. These deformed ears may be large and flabby and resemble Potter's description (suggestive of "Jumbo" or "Dumbo" ears), the ears might be folded over with the helix squared across its upper margin and bulbous, or they might be folded forward to mimic the cockle shell and be quite small, or they might be elfin-shaped. Lesser deformities of the ears are usually associated with marked asymmetry of the two ears.

The genito-urinary tracts in the 23 cases reported below came under suspicion because of the presence of one or more malformed ears, with the exception of Cases 19, 20, and 21, which were the first three encountered and are mentioned above. Cases 22 and 23 were of interest, as the ears resembled those of Cases 19, 20, and 21, but on one side only. At necropsy in both cases the kidney was absent on the same side. This necropsy finding drew attention to the possibility that this complex might be present in surviving patients.

Case Reports: Group A

Group A cases were four in which deformed ears drew attention to the underlying genito-urinary malformations, and in which the family histories revealed a high incidence of malformed ears and/or congenital anomalies of the genito-urinary tract in each of three generations (Table I).

TABLE I.—*Group A. Four Children Whose Malformed Renal Tracts Were Suspected Because of Malformed Ears; In Each Case the Father was Unexpectedly Found to Have Deformed Ears, Leading to a Search Into the Familial Incidence*

Case No.	Age (in Years) and Sex	Presenting Clinical Picture	Genito-urinary Malformation	Family Members Affected in Generations I, II, and III
1	9 M	Headache, pyrexia, colic, pyuria, grossly deformed bat-ears	Bilateral double ureters	I (1) Grandfather (ears + kidneys) II (2) Father (ears + kidney) II (3) Father's brother (ears + kidney) III (4) Brother (ears + raised blood urea)
2	4 M	Pyrexia, grossly deformed ears	Bilateral double ureters	I (5) Grandmother (ears + kidney) II (6) Father (ears + kidney) II (7) Father's sister (ears + kidney) III (8) Cousin (ears + kidney)
3	Birth M	Moribund newborn infant with typical facies of renal agenesis	No kidneys	I (9) Grandmother (kidney) II (10) Father (ear + hypospadias) III Cousins (11) Absent left kidney (12) Bat-ear + hypospadias (13) ,, ,,
4	6 M	Moribund, widespread neoplastic disease, deformed left bat-ear	L. cystic kidney	I (14) Grandfather (polycystic kidney) II (15) Father (L. polycystic kidney + ear) II (16) Father's sister (L. polycystic kidney + ear) III (17) Cousin died (cystic kidney) III (18) Brother (L. polycystic kidney)

Case 1

A boy aged 9 years was referred from another hospital, where the cause of a current pyrexia could not be determined. (He was a paratyphoid contact, and the following investigations had proved normal: urinalysis, tuberculin sensitivities, serum agglutinations against salmonella organisms, Wassermann reaction, and stool cultures. Headache, colic, and pyrexia had been persistent. His ears were so ugly and so similar to those infants with renal agenesis that a pyelogram was made. This demonstrated bilateral bifid ureters. His fever was due to an *E. coli* pyelitis. Parental interview disclosed that his father had the identical ear pattern and had been treated for two years for lumbar fibrosis. His urine was found to be infected, and pyelography showed double ureters. Father and son looked very similar; they both had receding chins, and were myopic but highly intelligent. The ears were folded forward at 90 degrees to the head, set low, bulbous, and ugly (similar to those shown in Fig. 2).

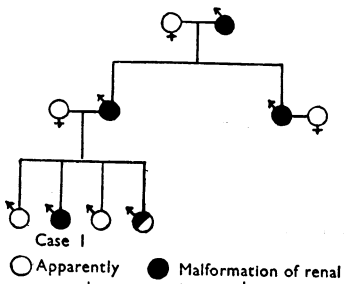

FIG. 1.—Family tree of Case 1.

A further family history (Fig. 1) disclosed the following. The paternal grandfather had such ugly ears that he would only be photographed in one profile. He had one kidney removed in adolescence for stones, and he died as a result of an operation to remove stones from the other one. In one paternal uncle with the same ear pattern renal disease was suspected. The youngest of the patient's three brothers, whose ears were similar, had an indeterminate pyelogram, but a blood urea of 55 mg. per 100 ml. The other two brothers had normal ears and pyelograms.

Case 2

A boy aged 4 years, convalescent from meningococcal meningitis, continued to have pyrexia though all treatment had been discontinued. He had such ugly ears (Fig. 2) that infection in a malformed renal tract was suggested as the cause of his fever. Pyuria was demonstrated, *E. coli* was grown from his urine, and his pyelogram demonstrated bilateral double ureters. His father had similar, if not quite as ugly, ears (see inset on Fig. 2). Pyelography was performed and displayed kinking with excessive filling of the upper end of the right ureter, together with bifid formation of the left renal pelvis.

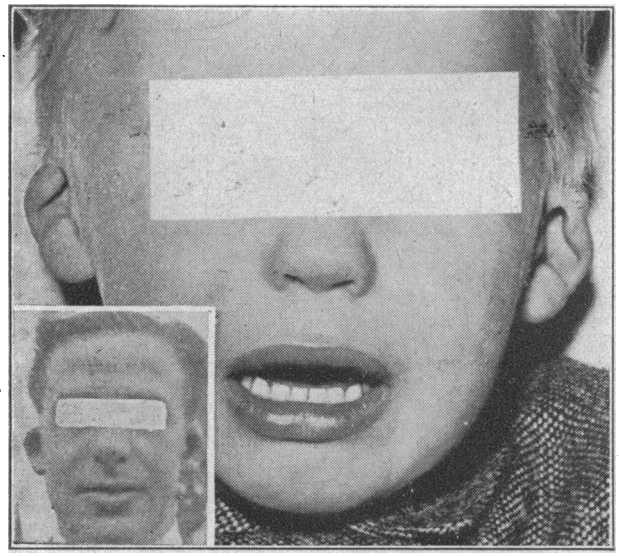

FIG. 2.—Case 2, with father inset. Note deformed ears. Similar ear patterns occurred in Case 1 and affected family members of Case 1 (see text) and in Case 21.

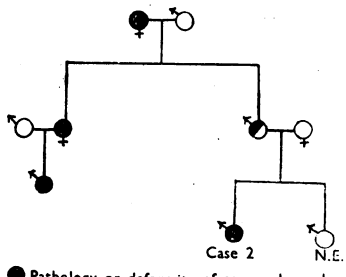

FIG. 3.—Family tree of Case 2.

A further family history (Fig. 3) was as follows. The paternal grandmother had similar ears and died while undergoing nephrectomy. A paternal aunt with renal disease died shortly after giving birth to a son, who in turn had the same ear pattern and was undergoing investigation for haematuria. The patient's brother has normal ears but has not had a pyelogram.

Case 3

A baby was born at full term, weighing 6 lb. 3 oz. (2,800 g.) with the typical facies of renal agenesis (see Fig. 4). The father was interviewed and noted to have a left flabby bat-ear. The infant died when 8 hours old, and at necropsy was found to have renal agenesis and hypospadias. The lungs were hypoplastic.

The family history and investigations produced the following information (Fig. 5): The father has a left bat-ear and a hypospadias ("hooded penis"). The paternal grandmother had died of "cystic kidney disease." The paternal aunt was normal, and pyelography was normal. She had had four pregnancies. One child had died unexpectedly at 6 years and was found to have only one kidney. Two of the children had left bat-ears and hypospadias but normal pyelograms, but in another the ears, penis, and pyelogram were normal.

This family is of interest not only because it shows the incidence of this clinical complex in each of three generations but also because it appears to show a genetic relationship between deformed ears, hypospadias, unilateral absence of a kidney, and absence of both kidneys. Potter (1952) states that, in her experience, there is no demonstrable association between unilateral and bilateral absence of kidneys.

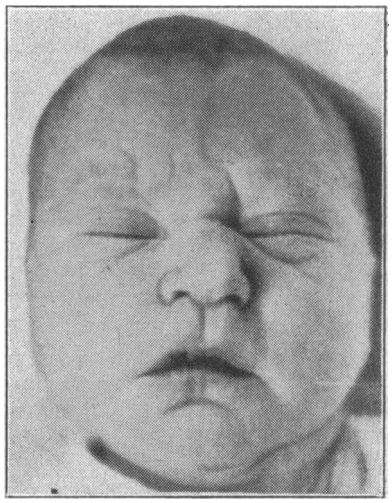

FIG. 4.—Case 3. Typical facies of renal agenesis. Here ears were "Dumbo" type as in Case 17 (see Fig. 11). For other published photographs see bibliography.

Case 4

Polycystic disease of the kidneys has a well-known familial incidence. However, no record could be found of an association of malformed ears with this familial trend.

A boy aged 6 years was transferred to a children's ward dying from generalized malignant disease. He was noted to have a low-set deformed left ear. His father also had a similar left ear, and, when challenged, stated that not only had he had a left-sided nephrectomy for polycystic disease of the kidney but his own father and one sister had died with this disease (Fig. 6). This sister had had two children, one of whom had died with diseased kidneys when a few hours old; the other had multiple congenital deformities, including microcephaly, but there was no knowledge about the state of the kidneys. The patient had one brother with cystic disease of his left kidney but with normal ears. At necropsy the patient was found to have widespread infiltrating neoplastic deposits and a left-sided polycystic kidney (which was also affected by the neoplasm).

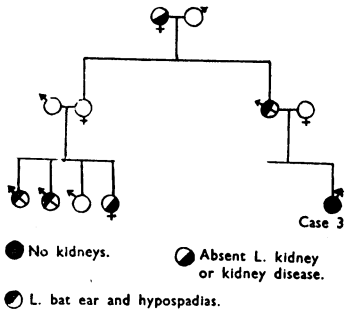

FIG. 5.—Family tree of Case 3.

Group B

In these cases the presence of bat-ears aroused suspicion of associated malformation of the renal tract being the basis of the pathology. In every case in which only one bat-ear was present it was the renal tract on the same side that proved to be abnormal. Family histories and investigations were incomplete (Table II).

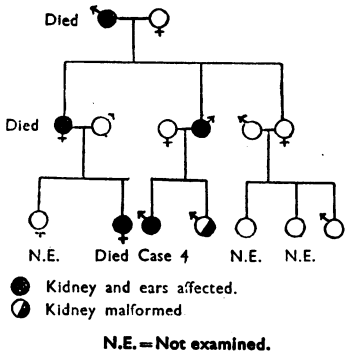

FIG. 6.—Family tree of Case 4.

Case 5.—A boy aged 8 years attended with a cough, sweating, and abdominal colic. He had a large mobile mass palpable in the left hypochondrium; this was thought to be an enlarged spleen, but as he had a gross left bat-ear a pyelogram was made. This showed a left double kidney.

TABLE II.—*Group B (Nine Cases)*

Case No.	Age and Sex	Presenting Clinical Picture	Ear Formation	Genito-urinary Malformation
5	8 M	Colic, sweating, cough	Left bat-ear	L. double ureter (heminephrectomy)
6	6 F	Enuresis	L. pinna squared off with bulbous upper margin (Fig. 8)	L. double ureter
7	9 M	Renal dwarf. Multiple fractures. Weight 39 lb. (17·7 kg.)	L. elfin-shaped bat-ear	Absent L. functioning renal tissue
8	4 M	Enuresis, dysuria	Bilateral asymmetrical bat-ears	R. hydronephrosis. L. bifid ureter
9	13 M	Colic 2 years	L. bat-ear	L. hydronephrosis. Plastic repair of L. ureter (Fig. 10)
10	3 M	Haematuria, colic, and dysuria for 3 days + palpable mass	R. bat-ear	R. hydronephrosis
11	9 M	Enuresis, dysuria	Bilateral bat-ears L. > R.	L. double ureter. L. heminephrectomy
12	11 M	Enuresis, colic	L. bat-ear	L. hydronephrosis
13	4 M	,, ,,	Bilateral bat-ears	Bilateral bifid ureters

He later developed recurrent pyelitis and haematuria, so a left heminephrectomy was performed (see Fig. 7).

Case 7.—A boy of 9 years with radiological and clinical evidence of renal rickets had multiple fractures. His blood pressure was 105/70; blood urea, 80 mg. per 100 ml.; alkaline phosphatase and serum phosphorus were raised, but blood calcium and renal urea clearance were diminished. Long bones showed radiological evidence of rickets. He had a left elfin-shaped bat-ear and two prograde pyelograms failed to show any renal tissue on the left. His palpable right kidney was normal on retrograde pyelography (see Fig. 9).

Case 10.—A boy of 3 years was admitted to hospital with a history of abdominal pain, haematuria, and dysuria for three days. He had a palpable tender mass in the right loin and a marked right deformed bat-ear. Pyelography and operation demonstrated a right-sided hydronephrosis.

Case 11.—A boy aged 9 years had been discharged from a hospital after recovering from pneumonia. He had bilateral bat-ears; the left one was more obvious, flabby, and folded over. He had pyuria, and pyelography demonstrated a left double ureter. A left heminephrectomy was performed.

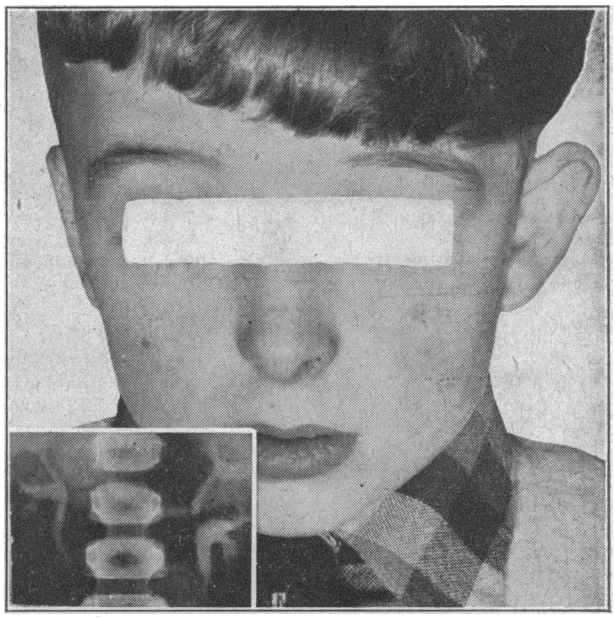

FIG. 7.—Case 5. Large left bat-ear, deficient in cartilage. Pyelography showed double left ureter. Similar ear formation in Cases 11, 12, and 13. Relatives of Case 3 had a similar left bat-ear.

Group C

In this group of 10 cases there were some infants in whom genito-urinary malformations were suspected before necropsy because of misshapen ears. Cases 19 and 20 (Table III) were the first seen in this series. Family histories were not investigated.

Discussion

Clear association occurred between malformations of the ears and the genito-urinary tract in the cases described. Sometimes the ear was grossly deformed; in others there was unilateral bat-ear, but in all there was asymmetry. Appreciation of the abnormal ear requires careful observation and an appreciation of the surprisingly constant forms of the normal ear.

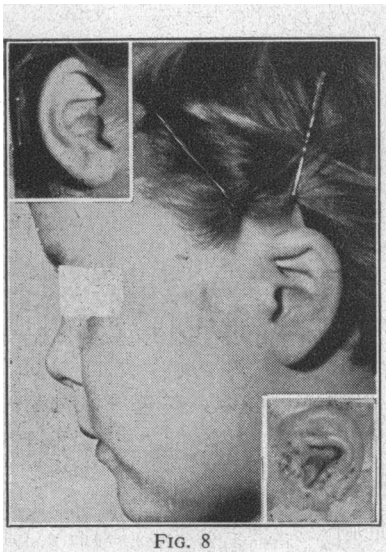

FIG. 8

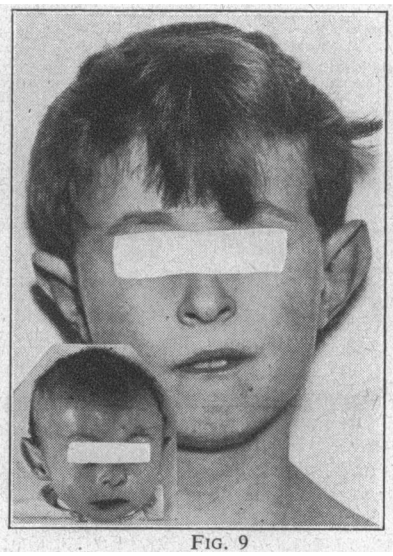

FIG. 9

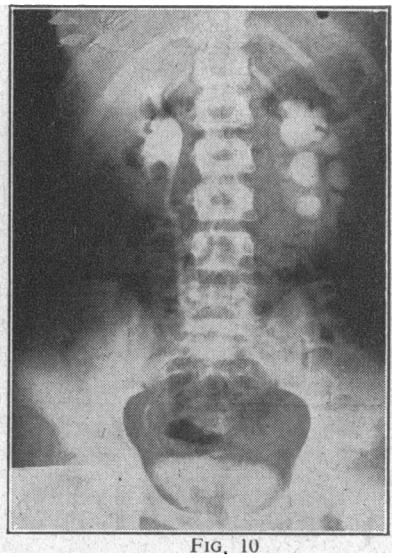

FIG. 10

FIG. 8.—Case 6. Normal right ear inset top left corner. Left pinna "squared off and bulbous." She has a left double ureter. Left ear of Case 15 is inset lower right corner to show similarity. Case 15 had cystic kidneys. Similar ears were displayed by Cases 18 and 21. FIG. 9.—Case 7. Renal rickets. Note left elfin-shaped bat-ear. No evidence of left functioning renal tissue. Inset infant, with similar right elfin ear, found at necropsy with cystic right kidney and other defects. Not described in text. Similar ear formation present in Cases 9 and 10. FIG. 10.—Case 9. Pyelogram. Had left bat-ear, and complained of colic two years. Ear similar to Case 7.

The patterns of abnormality encountered were deficiency of cartilage in large or small flabby ears, folding over of the upper ear, small folded cockle-shell ears, and ears that are "squared off" across the superior margin of the helix with a bulbous thickening of the flat upper edge of the helix. As the children have grown older many of the deformities have become less obvious.

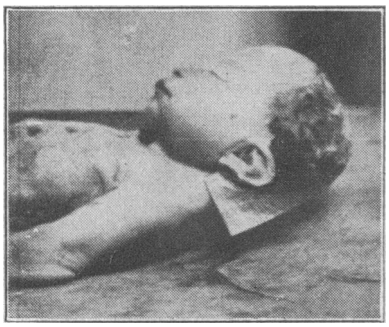

FIG. 11.—Case 17. Large asymmetrical "Dumbo" ears. Absent left kidney. Similar ear formation was noted in Cases 1, 14, 19, 20, and 23 (see text).

Incidence

In the period under review 23 cases were encountered together with 18 relatives involved with this syndrome. Four of the children seen had deformed pinnae and enuresis or pyelitis, but pyelography proved normal. In addition one infant had no external left auditory canal and only a vestigial left pinna; the pyelogram showed bilateral secreting renal tissue. However, in the same period three children encountered had normal ears but were found to have double ureters. Large numbers of pyelograms have been made for numerous reasons other than malformed ears, but only in the three cases mentioned has renal tract abnormality been found with normal ear formation.

Eight cases of renal agenesis (not all reported here) were recognized in approximately 18,000 births covering six years. Davidson and Ross (1954) found 5 cases in 368 consecutive necropsies on newborn infants.

Barclay and Baird (1935) examined 500 consecutive pyelograms and found six cases of bifid ureters. Dr. Fawcitt, of Booth Hall Children's Hospital, kindly examined consecutive pyelograms of 50 children and 50 adults and found only one case of a bifid ureter—approximately the same incidence—but this was in a series of cases referred for pyelography because renal tract abnormality had been suspected on clinical grounds (without reference to the ears). This paper reports 19 cases subjected to pyelography because the presence of peculiar ears suggested that the clinical symptoms were associated with anomalies of the renal tract. Only four did not have such anomalies displayed. A further 11 infants who came to necropsy demonstrated this close association, but no case is recalled of an infant coming to necropsy who did not have genito-urinary malformation when he had malformation of the pinnae. Finally, the 18 relatives listed in Table I were investigated for abnormalities of the renal tract because one member of the families had peculiar ears and associated renal anomalies.

Aetiology

Most authors have expressed the view that renal agenesis, absence of one kidney, and double ureter formation are unrelated. Certainly renal agenesis has been thought to be most probably due to some noxious influence occurring at a crucial stage in the development of the foetus, but animal experiments suggest a clear genetic association (Davidson and Ross, 1954). It now appears that, in some cases at least, all these conditions are interrelated and genetically determined, but not sex-linked.

Of the pathological conditions mentioned in this paper, only polycystic disease of the kidney seems to have a well-recognized familial incidence. Madisson (1934) described the case of a mother who gave birth to two infants with renal agenesis, but thought that this was due to mutation causing a defect in the germ plasm. No other case has been found in a search of the literature.

TABLE III.—*Group C (Ten Cases)*

Case No.	Weight	Estimated Gestation in Weeks	Age at Death	Ear Formation	Genito-urinary Malformation	Other Malformation and Comment
14	3 lb. 10 oz. (1,640 g.)	34	3 days	Very large flabby ears. "Dumbo ears"	Rudimentary penis, hypospadias. No scrotum	
15	4 lb. 5 oz. (1,950 g.)	30	4 hours	Small asymmetric folded ears	Bilateral cystic kidneys	Anal atresia. Microphthalmos. Omphalocele
16	4 lb. 10 oz. (2,100 g.)	37	10 mins.	Small deformed folded ears set low	Minute kidneys, R. smaller than L. Bifid ureters	Oesophageal atresia. Hypoplastic lungs
17		28	59 hours	Very large flabby "Dumbo ears" set very low (Fig. 11)	Absent left kidney	Hypoplastic atelectatic lung. Very large adrenal glands
18	4 lb. 8 oz. (2,040 g.)	25	9 ,,	Small folded ears. R. much smaller than L. ear. R. ear squared off across top	R. kidney ½ size of L. R. ureter ended blindly in posterior bladder wall. L. ureter had 3 partial stenoses or valves, with intervening dilatations	Large patent foramen ovale and ductus arteriosus
19	3 lb. 7 oz. (1,560 g.)	32	5 mins.	Bilateral flabby low-set "Dumbo ears"	No kidneys	Typical Potter facies
20	4 lb. 3 oz. (1,900 g.)	40	Stillborn	Bilateral large flabby low-set ears	No kidneys. Small phallus, labia majora. Sex indeterminate	Typical facies. Anal atresia
21	3 lb. 10 oz. (1,640 g.)	32	4 hours	Bilateral small ears, low set, asymmetrical	No kidneys or ureters. Male organ formation	Facies very similar to Potter's infants, but small folded ugly ears
22	7 lb. 11 oz. (3,490 g.)	39	6 days	R. ear small, squared off folded and low set	Absent R. ureter and kidney. L. kidney in pelvis. No urethra. Male	
23	3 lb. 13 oz. (1,730 g.)	36	20 hours	Flabby, large R. low-set ear with upper edge squared off	Male. Absent R. kidney. L. kidney in pelvis	Oesophageal atresia. Malrotated gut

Summary

Attention is drawn to the association of deformed ears and congenital malformation of the genito-urinary tract.

The ear deformities are similar to those seen in infants with renal agenesis.

Not all infants who die with renal agenesis have the large flabby ear described by Potter, but other patterns occur and are mentioned.

There seems to be a genetic association between renal agenesis, unilateral absence of one kidney, peculiarity in the formation of the ears, and other malformations of the genito-urinary tracts.

A malformed ear in the presence of enuresis, dysuria, abdominal colic, or other symptom referable to the genito-urinary tract justifies early pyelography.

The familial association of this clinical complex does not appear to be sex-linked.

A number of cases of unilateral bat-ear deformity are described in which the renal tract on the same side as the abnormal ear was affected. This might be co-incidental.

Congenital anomalies of the genito-urinary tract occur without associated defects of the ears. In this series this has been in the ratio of 3 : 41.

Deformity of the ears can occur without deformity of the genito-urinary tract.

Appreciation must be expressed to good-humoured colleagues, secretaries, and clinical photographer Miss Stanley. Particular thanks are due to those allowing me to utilize their material and reports, particularly to the medical staff of the Liverpool Maternity Hospital; to Professor Capon, Drs. Rewell, Parry Jones, Adderley, and Jackson for necropsy reports; to Drs. Fawcitt, Livshin, Taylor, and Walton for radiological findings; to Mr. Grime for Cases 9 and 10; and to Professor W. Gaisford for his invaluable help in the preparation of this paper.

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PACHYDERMO-PERIOSTOSIS (IDIOPATHIC OSTEOARTHROPATHY)

BY

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It has long been recognized by physicians that clubbing of the fingers may occasionally occur in healthy people. In 1935 Touraine, Solente, and Golé pointed out that, in some patients in whom no evidence of intrathoracic or cardiac disease could be found, gross clubbing was associated with enlargement of the extremities, thickening of the skin, particularly over the face, and a periosteal reaction in the long bones. They proposed the term "pachydermie plicaturée avec pachy-periostose des extrémités" for the condition. In 1941 Brugsch showed from a study of the literature that a hereditary element was involved. In all, some 50 cases of this syndrome have now been described, mostly in the French literature.

Examples of the condition have been described under many different names—for example, megalis cutis et ossium (Gronberg, 1927), generalized osteophytosis (Freund, 1938), hyperostosis generalisata with pachydermia (Uehlinger, 1941, and most German authors following him). Most recent authors (Vague, 1948; Lièvre *et al.*, 1949; Franceschetti *et al.*, 1950; de Sèze and Jurmand, 1950; Findlay and Oosthuizen, 1951) use the term pachydermo-periostosis, which has the twin merits of brevity and accuracy.

So far as I know, no case has been described in England, and I therefore present three cases in order to draw attention to the syndrome.

Case 1

A single man aged 31 was admitted to University College Hospital on October 28, 1951, because of a sudden attack of pain in the left lower chest associated with "giddiness." The pain cleared up spontaneously over the next few hours. His past history had been eventful. At the age of 4 he had been an in-patient at Great Ormond Street, where he was treated for swelling of the knees. He also had genu valgum, for which he was in plaster for about 15 months, without improvement. No further details are available about this admission. At the age of 6 he first began to have attacks of loss of consciousness, with or without pain. Since adolescence he had suffered from very heavy sweating, particularly from his hands and feet; at one time he used to rot through a pair of boots every three months. During 1942 and 1943 bilateral cervical and lumbar sympathectomies were carried out. At that time it was noticed that his hands and feet were unusually large. After these operations the attacks of giddiness and pain were reduced in frequency, and the amount of perspiration was greatly diminished. In the subsequent years he developed varicose veins in both legs and suffered from numerous attacks of thrombophlebitis. A number of operations were carried out on his legs; the veins were first tied, and later removed. He still complained of considerable pain in the legs at the time of his admission.

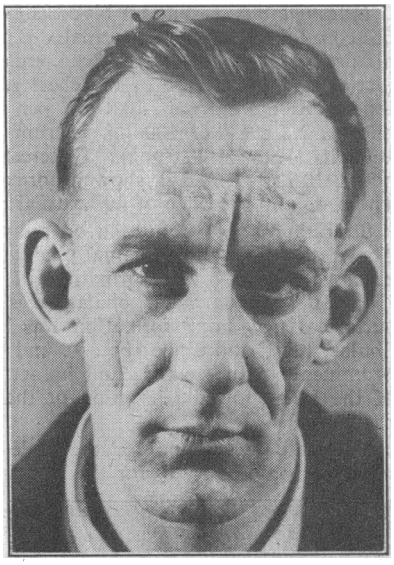

FIG. 1.—Case 1. Note the deep furrows on the face and above the eyebrows.

He thought that his hands and feet had always been large. He did not think that there had been any sudden increase in size at or about puberty. None of his family had large hands or feet.

On examination he was a tall thin man. His face was prematurely aged and was marked by very prominent skin folds on the cheeks and between the brows, and by exaggeration of the normal nasolabial folds (Fig. 1). His nose was large, shiny, and bulbous. The supraorbital ridges were prominent. The skin of the face was thickened and inelastic, and there was considerable sebaceous secretion. There was no thickening of the scalp. Neither the jaw nor the tongue was enlarged. His height was 72 in. (185 cm.); head to pubis 34½ in. (87.5 cm.), pubis to ground 38½ in. (98 cm.). The span of the outstretched arms was 74 in. (188 cm.).

Apart from the facies, the most striking feature was the gross enlargement of the hands and feet. Both wrists

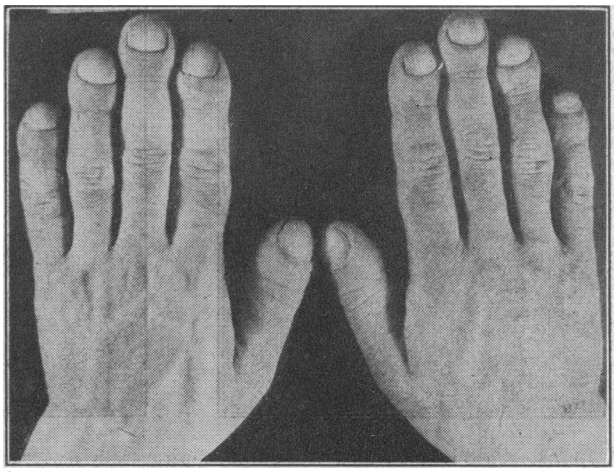

FIG. 2.—Case 1. Enlargement of hands and clubbing of fingers.