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CONGENITAL DEFICIENCY
OF
ABDOMINAL MUSCULATURE
WITH ASSOCIATED UROGENITAL MALFORMATIONS

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INTRODUCTION

Congenital deficiency of the abdominal musculature associated with urogenital anomalies is a comparatively rare pathological syndrome occurring almost exclusively in males. The typical complex consists of:

1. Deficiency of abdominal musculature particularly below the umbilicus.
2. Hypertrophied urinary bladder, usually accompanied by hydro-ureter and hydronephrosis.
3. Often a malrotation of the gastro-intestinal tract.
4. Pigeon breast deformity of the thorax.
5. Cryptorchism.

The consistency of this particular combination of abnormalities in the cases presented here has given rise to much investigative effort to determine the pathologic embryology.

The purpose of this paper is to report one new case presenting this syndrome and to review fifty-three more from the literature. The various theories of pathogenesis will be discussed.

CASE REPORT[45]

J. G., a white male, age three months, was admitted to Children's Memorial Hospital, Omaha, Nebraska, with the complaint of an "enlarged abdomen with poor muscle tone."

Present Illness:

The baby was noted at birth to have an enlarged abdomen, the anterior wall of which was thrown into folds like loose skin. A flat X-ray of the abdomen at this time failed to reveal pathology.

Since birth the mother has noticed that the baby appeared pale and seemed to be generally weak. The bladder was often seen extending to the level of the umbilicus. He cried with urination and defecation and seemed to pass unusually large amounts of urine each time. Stools had been soft and pasty, but an enema had been required on alternate days for the past month.

Birth History:

The patient was the first child of healthy young parents. The mother had a normal pregnancy. Labor was twelve hours with a cephalic occiput posterior presentation. Ether anesthesia was given in the second stage and a mid-forceps delivery accomplished. The baby was cyanotic and cried weakly, so was placed in oxygen for the first two days of life.

Development:

Birth weight was seven pounds, fifteen ounces, and weight at three months was eleven pounds, four ounces, a gain of about one pound per month.

He was able to hold his head up at two months and could, at three months, roll over.

Family History: Negative.

Physical Examination:

This revealed a poorly developed, fairly well-nourished, white male of three months who appeared quite pale.

Head: There was overriding of the occipital and parietal sutures.

E.E.N.T.: No pathology noted.

Thorax: Flattened in the anterior-posterior diameter with a prominence and increased angulation of the costalchondral junctions. No pathology discovered in heart or lungs.

Abdomen: Skin was wrinkled and loose, suggesting great distention at some previous time. Abdominal wall was very thin and loops of bowel could be palpated throughout. The urinary bladder was visible and palpable at the umbilicus, but this disappeared after voiding. There was a bulging in the right flank which became more pronounced when intra-abdominal pressure was increased. Rectus muscles were present bilaterally, but were poorly developed.

Genitalia: Testes were not palpable in the scrotum or in either canal.

Back: Musculature appeared underdeveloped.

Extremities: Muscles were hypotonic.

Hospital Course: Patient was in the hospital about eleven weeks. During that time he spiked a temperature ranging from 101° to 104° F. nearly every day without apparent signs of infection except for one urinalysis, which showed numerous bacteria and pus cells. Urinalyses were otherwise negative, except for a consistently low specific gravity (1.001 - 1.005). Blood NPN was elevated with a range of 43.7 mgm. % to 86.0 mgm. %. A cystogram showed a large bladder with reflux of the opaque material into an elongated, tortuous left ureter and a markedly dilated left kidney pelvis. Right ureter was not visualized. The bladder was sharply cut off at the urethral meatus, suggesting to the radiologist, a congenital valve obstruction. On cystoscopy, a #10 infant cystoscope was passed with no sign of obstruction. The ureteral orifices could not be identified even with the aid of indigo carmine injected intravenously.

An intramuscular pyelogram showed a rapid absorption of diodrast, but insufficient excretion for visualization of the material in any part of the urinary tract.

A suprapubic cystostomy was performed to relieve urinary retention, and the bladder was found to be very large with a wall about one-fourth inch thick. A #20 mushroom catheter was left in place, but the blood NPN failed to change significantly in the postoperative period.

Treatment in the hospital consisted essentially of penicillin for the control of infection and of the permanent

catheter drainage mentioned above.

He was discharged with his condition unchanged and a guarded prognosis.

Progress Note: Patient was followed by Dr. J. Harry Murphy as an office patient. He continued to be a happy child, but was slow mentally and failed to grow.

When last seen at eleven and one-half months of age, he weighed thirteen pounds, three and one-half ounces.

The suprapubic catheter was still in place and there had been no evidence of urinary infection.

Future plans include hospitalization for establishment of urethral drainage, and determination of the status of the right kidney.

SUMMARY C

Author	Age	Sex	Abdominal Muscles	Testes
1. Henderson[3]	60 yrs.	M	Lower half of rectus absent. Upper half well developed; others deficient.	Undescended
2. Parker[4]	Newborn	M	Very rudimentary. Transversus absent.	Undescended
3. Guthrie[5]	9 wks.	M	Costal margins of all muscles present. Rest absent.	
4. Platt[6]	2 yrs.	M	Transversus absent. Rest present, but deficient.	Undescended
5. Osler[7]	6 yrs.	M	Upper half of rectus present. Rest absent.	Undescended
6. Stumme[8]	17 yrs.	M	Transversus and upper half of rectus present. Rest absent of deficient.	Undescended
7. Garrod & Davies[9]	8 wks.	M	All muscles deficient. Strands of upper rectus present.	Undescended
8. Bolton[10]	3 wks.	M	Rectus present in upper half. Oblique muscles present only at costal margin.	Undescended
9. Hall[11]	2 wks.	M	Lower rectus absent. Rest of muscles very deficient.	Undescended
10. Pels-Leusden[12]	3 mo.	M	Trace of rectus present. Rest absent.	Undescended
11. Pels-Leusden[12]	7 wks.	M	Rectus present. Others unmentioned.	Undescended

REPORTED CASES

Thorax	Urogenital Tract	Progress	Treatment
	Hydronephrosis & hydro-ureter. Bladder hyper-trophied. No obstruc-tion.	Died of collapse and emphysema.	
Pigeon breast	Bladder dilated and adherent to umbilicus. Ureters dilated. No obstruction.	Died of a wasting process. Opisthotonos, but no convulsions.	
Deformed	Bladder & kidneys not palpable.	Living at time of report.	
Harrison's groove and protrusion of sternum.	Kidneys palpably enlarged. Bladder extends to um-bilicus.	Living at time of report.	
Harrison's Groove	Bladder large & adherent to umbilicus. Ureters dilated. Right kidney enlarged. No urethral obstruction.	Living at time of report.	
	Left kidney and ureter dilated.	Died of erysipelas and broncho-pneumonia.	
	Bladder dilated and hypertrophied. Phimosis.	Died of furunculosis and suppurative pericarditis.	Circumcised for phimosis.
	Ureters and kidneys are dilated. Bladder hyper-trophied. Patent urachus.	Died suddenly. Necropsy did not determine cause.	
Pigeon breast	Bladder hypertrophied. Right kidney small. Ureters dilated and tortuous. No urethral obstruction.	Died of peritonitis. Post-mortem done.	
Flattened laterally.		Living at time of report.	

Author	Age	Sex	Abdominal Muscles	Testes
12. Levy[13]	70 yrs.	M	Lower half of rectus absent. Obliquus internus present on right, absent on left.	Undescended
13. Mollison,[14] Mackeith and others[15]	2 yrs.	M	Upper rectus partly present. Rest absent.	Undescended
14. Smith[16]	2 mo.	M	All muscles absent.	
15. Thatcher[17]	3 wks.	M	A few upper rectus fibers present. Rest are absent.	Undescended
16. Stern[18]	6 wks.	?	All muscles apparently absent.	
17. Eckhoff[19]	10 days	M	Upper rectus present. Rest absent or very deficient.	Undescended
18. Blasi[20]	2½ mo.	M	Rectus partially present. Rest absent.	
19. Ikeda and Stoesser[21]	5 mo.	M	All muscle layers absent.	Undescended
20. Taillens[22]	2½ mo.	M	Transversus absent. Rest partially present.	
21. Brindeau and Jacquet[23]	2 mo.	M	Rectus present. Rest absent.	Undescended
22. Poli[24]	3 mo.	M	No response to electrical stimulation.	Undescended
23. Hofstein[25]	Newborn	F	No response to electrical stimulation.	
24. Moncrieff[26]	6 mo.	M	A few strands of rectus. Rest absent.	Undescended
25. Malossi and Gelli[27]	10 mos.	M	Trace of muscle in upper rectus. Rest absent.	

Thorax	Urogenital Tract	Progress	Treatment
		Living at time of report.	
Harrison's Groove.	Normal at first. Developed a dilated U.G. system at age of 12 with symptoms of uremia.	Living and functioning fairly well at age of 12 years.	
	Bladder palpable above pubis when empty.	Living at time of report.	
Pigeon breast	Bladder and ureters dilated. Right hydronephrosis. No urethral obstruction.	Apparently did well, but died suddenly of bronchopneumonia at 2 months.	
	Bladder hypertrophied.	Living at time of report.	
Narrowed. Harrison's Groove.	Bladder hypertrophied. Hydro-ureters & hydronephrosis. No obstruction.	Blood-streaked urine. Pyrexia, diarrhea, and wasting. Died at age of 9 weeks.	
Narrowed.	Right kidney palpable.	Living at time of report.	
Pigeon breast	Bladder hypertrophied and adherent to umbilicus.	Died of pneumonia.	
		Died of diarrhea and malnutrition.	
Narrowed. Prominent.		Living at time of report.	
Normal	Kidneys palpable. Bladder hypertrophied.	Living at time of report.	
Elevated		Living at time of report.	
Deformed	Hydro-ureter and hydronephrosis. Bladder adherent to abdominal wall. No urethral obstruction.	Died of pneumonia.	
Elevated	Hydronephrosis and hydroureter. Bladder hypertrophied. No urethral obstruction.	Died of respiratory infection.	

Author	Age	Sex	Abdominal Muscles	Testes
26. Baxter[28]	4 wks.	M	No evidence of any abdominal muscle by physical exam.	Undescended
27. Baxter[28]	17 yrs.	M	No evidence of any abdominal muscle by physical exam.	
28. Baxter[28]	6 wks.	M	Left transverse and oblique muscles defective. Rest intact.	
29. Friedley[29]	1 yr.	M	Few muscle fibers present at autopsy.	
30. Stoesser[30]	Newborn	M	Defective.	Undescended
31. McClendon[31]	6 mo.	M	A few fibers of left transverse present. Rest absent.	
32. Housden[32]	Newborn	M	External oblique present in part. Rest absent.	Undescended
33. Gibbens[33]	2 mo.	M	All muscles absent.	Undescended
34. Smith[34]	3 mo.		No muscle tissue palpable.	
35. Lichtenstein[35]	3 yrs.	M	No muscle present at autopsy.	Undescended
36. Howard[36]	Newborn	M	All abdominal muscles are deficient.	Undescended
37. Howard[36]	Newborn	M	No evidence of abdominal musculature.	Undescended

Thorax	Urogenital Tract	Progress	Treatment
Lower border flared. Sternum protrudes.	Bladder hypertrophied.	Died of pneumonia at age of 6 months. No necropsy.	
ibs & sternum flare upward.	Bladder hypertrophied. Tenderness over right kidney.	Recurrent pyelonephritis since age 2. Prognosis poor.	
		Living. Prognosis good.	
	Bladder hypertrophied. Hydro-ureter & hydronephrosis. Valve obstruction in both ureters.	Died of uremia.	
Slight pigeon breast	Bladder hypertrophied.	Died of exfoliative dermatitis at age of 12 days.	
	Dilated left kidney and ureter. Bladder dilated. Pyuria and hematuria at times.	Died of septicemia.	
Pigeon breast	Hydronephrosis & hydroureters. Bladder was enlarged. Prostatic hypertrophy.	Died of uremia.	
	Bladder & ureters dilated. Kidneys normal. No urethral obstruction.	Died of diarrhea.	
	Bladder not hypertrophied, to palpation.	Died at age of 4 months. Cause unknown.	
Pigeon breast	Bilateral hydro-ureter and hydronephrosis. Bladder hypertrophied. No obstruction.	Broncho-pneumonia and peritonitis.	
	Bladder was dilated and hypertrophied. Atresia of ureters & urethral Kidneys were normal.	Died. Cause unknown.	
	Kidneys showed cystic degeneration. Ureters dilated above an atresia. Bladder hypertrophied.	Died. Cause unknown.	

Author	Age	Sex	Abdominal Muscles	Testes
38. Aldrich[37]	5 wks.	M	No abdominal musculature detected except for transversalis.	Undescended
39. Dout and others[38]	2 wks.	M	Thin, flacid abdominal wall seemingly devoid of muscle.	
40. Irvin and Kraus[39]	2 wks.	M	No muscle tissue present.	
41. Obrinsky[40]	5½ mo.	M	Rectus present. Other muscles poorly developed.	Undescended
42. Silverman & Huany[41]	Newborn	F	All muscles deficient.	
43. "	4 days	M	Upper rectus present. Rest absent.	Undescended
44. "	Newborn	M	Upper rectus present. Rest absent.	Undescended
45. Eagle and Barrett[42]	3½ mo.	M	Laxness of abdominal wall.	Undescended
46. "	11 mo.	M	Deficiency of all muscles, but reflexes present in all quadrants.	Undescended
47. "	Newborn	M	Laxness of abdominal wall.	Undescended
48. "	15 mo.	M	Lax abdomen.	Undescended

Thorax	Urogenital Tract	Progress	Treatment
	Hydro-ureter & hydronephrosis. Bladder hypertrophied. Urethral diverticulum.	Died of malnutrition.	
	Hydronephrosis. Dilated bladder. Large median bar, not apparently obstructing.	Constant urinary infection until treated. Doing well at time of report.	Transurethral resection of bar.
	Hydronephrosis & hydroureter. Bladder enlarged. Urethra patent, but narrow.	Died at age of 5 wks. from broncho-pneumonia and cystitis.	Suprapubic cystostomy
Narrow and short.	Left polycystic kidney. Both ureters dilated. Bladder hypertrophied. Urethral diverticulum.	Died of urinary infection and uremia.	Massive doses of penicillin.
Costal margin flared.	No symptoms. Appeared normal at surgery.	Operated for volvulus. Doing well at time of report.	
Normal	Hydro-ureters and hydronephrosis. Bladder attached to abdominal wall. No obstruction.	Recurrent kidney infection. Doing well at time of report.	Chemotherapy. Surgical repair of hydro-ureters.
	Bladder, ureters, and kidneys were dilated. No obstruction.	Died post-op. from cystic ureter interferring with respiration.	Resection of part of ureter and chemotherapy.
Normal	Bilateral hydronephrosis and hydro-ureters.	Having intermittent episodes of pyelonephritis with elevated NPN.	Antibiotics.
Normal	Bilateral hydronephrosis and hydro-ureters.	Intermittent hematuria & persistent pyuria. Acute obstruction of left ureter. Did well after surgery.	Symptomatic & surgical repair of both ureters.
Normal	Bilateral hydronephrosis and hydro-ureters. Hypertrophied bladder.	Retrograde pyelography showed marked improvement in hydroureters after surgery. Kidney function nearly normal.	Surgical repair of both ureters. Antibiotics.
Pigeon breast	Bilateral hydronephrosis and hydro-ureters. Fibrous bar obstructing bladder neck.	Several episodes of pyelonephritis. Retention continued after treatment because of atonic bladder.	Surgical repair of both ureters. Transurethral resection of bar.

Author	Age	Sex	Abdominal Muscles	Testes
49. Eagle and Barrett[42]	2 yrs.	M	Relaxed, distended abdomen.	Undescended
50. "	1 wk.	M	A loose asymetric sac. Muscle present at origins and insertions.	Undescended
51. "	5 wks.	M	Strands of muscle at costal border.	Undescended
52. "	Newborn	M	All muscles present, but deficient.	Undescended
53. "	Newborn	M	Apparently no supporting musculature.	Descended. Bilateral hydroceles.
54. Murphy[45]	3 mo.	M	Recti present, but debilitated. Rest clinically absent.	Undescended

Thorax	Urogenital Tract	Progress	Treatment
Flared lower border.	Hypoplastic left kidney. Right kidney and ureter dilated. Median bar fibrosis causing obstruction.	Intermittent pyelonephritis. Did well after treatment.	Transurethral resection of median bar. Antibiotics.
Normal	Enlarged bladder with cystic urachus. Pouch of bladder in such a position that urethra might be compressed. Hydro-ureter and hydronephrosis.	I.V. pyelogram showed no excretion. Urinary tract infection. Died of infection and uremia.	None
	Hydronephrosis and hydroureter. Bladder enlarged and perforated in infectious ulcer.	Persistent patency and infection of urachus. Urethritis. Died of Staph. peritonitis.	Urethral dilation allowed closure of urachus.
	Complete obstruction of urethra. Hydronephrosis and hydro-ureter. Hypertrophied bladder. Patent urachus.	Breech delivery. Baby died of intracranial hemorrhage.	None.
	Hydronephrosis and hydroureter. Urethral obstruction at the verumontanum.	Stillborn. Separation of cervical vertebrae in difficult delivery.	None.
lat in AP diameter. Costal-chondral junction flared.	Left hydronephrosis and hydro-ureter. Bladder distended & hypertrophied. Apparently a urethral stricture by X-ray.	NPN varried from 40 to 86 mgm. %. Progressing well after treatment, but wt. gain has been slow.	Cystoscopy. Suprapubic cystostomy.

SUMMARY OF THE CASES

Number of Cases:

Fifty-four cases are considered here including the one seen by the author. Two early reports by Frolich,[1] in 1839, and von Ammon,[2] in 1842, have been omitted because information was insufficient for a diagnosis. Likewise, a case reported by Parsons and Bailing,[46] in 1933, is not considered because information was too scant to contribute to the knowledge of the syndrome.

Sex:

Fifty were male, two were female, and in two cases the sex was unmentioned.

Progress:

Thirty of the fifty-four patients died before the age of two years. The predominant causes of death were pneumonia, mentioned eight times, and kidney infection with uremia, given in four cases.

The remaining twenty-four were living at the time of the report. One was sixty years and another seventy years of age. These two cases, reported by Henderson[3] and Levy,[13] apparently had little urogenital pathology. Patients handled by Stumme[8] and Baxter[28] were both seventeen years of age, but prognosis for long life was poor because of kidney involvement.

The other twenty living cases were under six years of age, most of them in their first year of life. Condition at time of report was omitted in many of the early cases, but Eagle and Barrett,[42] who

have five patients living, report that after surgical alleviation of urinary obstruction and control of infection by antibiotics, they were able to increase kidney function and offer a good prognosis.

Family History:

No tendency for fetal anomalies or other unusual feature has been noticed.

Obstetrical History:

There is nothing remarkable in the obstetrical history; the great majority of the cases are products of normal gestations and deliveries.

Abdominal Muscles:

No case was found where thorough investigation revealed a complete absence of abdominal musculature. The rectus abdominus was most consistently present, usually being best developed in its upper portion. The external oblique was encountered slightly less often while the transversus and the internal oblique may be grouped with the lower half of the rectus as the most frequently deficient. Throughout the study of the cases, a tendency was noted for the persistence of strands of muscle tissue along the costal margins and at the lateral pelvic brim.

Urogenital System:

The bladder was dilated and hypertrophied in thirty-eight cases. Five patients were reported to have had normal bladders; however, in only two of these was the condition proved by cystogram, surgery, or autopsy. The other three had bladders not palpably

enlarged, but one of these went on to develop dilation with signs of uremia twelve years later.[14] The condition of the bladder was not mentioned in the other eleven reports. In eleven patients the bladder was described as adherent to the umbilicus or anterior abdominal wall, while a patent or cystic urachus was reported three times.

Hydro-ureter, usually bilateral, was present in thirty-two cases, including every autopsied case except one. Obstruction due to kinking or atresia of a ureter was common.[42]

Hydronephrosis, also usually bilateral, was present in thirty cases; in four patients one kidney was atrophic and there were two with polycystic kidneys. Some form of kidney pathology was demonstrated in every autopsied case except two.

The urethra was described as unobstructed in fourteen cases. Fibrosis of median bar, diverticulum, prostatic hypertrophy, complete atresia, or some other form of obstruction was described in nine patients. Since Housden[32] discovered the first urethral obstruction in 1934, and emphasized the importance of looking for it, this condition has been discovered in forty percent of the cases.

Testes:

Of the fifty males being considered, the testes were undescended in thirty-nine. In one case belonging to Eagle and Barrett,[42] they were descended, but bilateral hydroceles were present. Position of the testes was not given in the other ten reports.

Thorax:

The thorax was described as deformed in twenty-five cases. The usual abnormalities were pigeon breast and Harrison's groove. In six cases the thorax was normal, but it is interesting that these six patients were very young at the time of the report.

Gastro-Intestinal System:

Anomalies of the gastro-intestinal system have occurred in ten cases:

1. Congenital megacolon with obstipation.(20)

2. Absence of mesoappendix, indicating a minor degree of malrotation, discovered incidentally at autopsy.(21)

3. Caecum had mesentery and lay on the left side of the abdomen. The appendix was anterior to the ileo-caecal junction. Discovered incidentally at autopsy.(26)

4. Caecum and appendix were near the midline below the left lobe of the liver. Caecum, appendix, ascending colon, and ileum had common mesentery.(35)

5. Stomach, appendix, and caecum were in the right upper quadrant while the ascending colon was doubled over and lay anterior to a normally placed transverse colon. Cause of death was given as diarrhea, pyrexia, and wasting.(19)

6. Mesocaecum was extremely long.(8)

7. Entire colon was suspended by a free mesentery. Discovered incidentally at autopsy.(39)

8. Patient developed signs of intestinal obstruction at

two to three days of age. At surgery a volvulus was found in the small bowel, and the caecum lay in the right epigastrium firmly attached to the posterior abdominal wall by fibrous bands. After surgery patient did well.(41)

9. Caecum and ascending colon were suspended by free mesentery. Discovered by barium enema and flouroscopy done for research purposes.(41)

10. Caecum incompletely rotated. Discovered incidentally at autopsy.(41)

Other Congenital Anomalies:

1. Talipes equinovarus was noted six times.

2. Webbing of all the toes was described once.(42)

3. Flexion deformity of right leg, genu valgum, and talipes calcaneo valgus.(16)

4. Quadratus lumborum muscle was deficient in four cases.

5. Congenital dislocation of the hips was present once.(17)

6. Hypospadius.(42)

PATHOGENESIS

Although congenital anomalies are frequently multiple, the fact that a dilated, hypertrophied bladder and probably hydro-ureter and hydronephrosis can be predicted, once the deficiency of abdominal musculature has been established, has led most observers to believe that the two conditions are causually related. Theories are as follows:

Abdominal Muscles.

1. There is a failure of these muscles to develop embryologically.[1,9,21,35]

The somites from which muscles develop appear in an embryo of about four or five weeks gestation. These somites, originally primitive mesenchymal tissue, differentiate in their lateral portions into myotomes or muscle plates. At about six weeks the myotomes, in the lower thoracic segments, fuse to form a single muscle column on either side of the developing spine. This muscle tissue grows ventrally and, due to a tangential splitting process, the transverse and oblique abdominal muscles are formed. When the ventral edge of this muscle sheet splits off longitudinally, we see the beginning of the rectus. This process is complete and definitive muscles are present at seven weeks.[44]

Now, according to the above theory, there is an arrest of development in this chain of events and, therefore, an absence of the muscles that are usually formed during this period. Facts that substantiate this theory are: (a) Serial sections at autopsy in two cases reveal large areas with neither muscle tissue nor fibrous remains of what might have been muscle tissue.[9,35] (b) In Mollison's[14] case the deficiency of abdominal musculature was present from birth while the dilation and hypertrophy of the urinary tract did not develop clinically until the age of twelve years.

Facts conflicting with the theory are: (a) There were scattered strands of normal muscle tissue in every case that was

thoroughly studied. They occurred chiefly at the costal margin and near the lateral brim of the pelvis. These are hard to explain on the basis of embryologic arrest. (b) There has never been, associated with this condition, a deficiency of intercostal muscles which develop in approximately the same way and at the same time.

2. Deficiency of abdominal musculature is secondary to urethral obstruction and pressure from a distended bladder. This could occur in two ways: (a) Muscles may be prevented from migrating ventrally. (b) Secondary atrophy of preformed muscle.

In considering these two possibilities, the time relation becomes important because, if a distended bladder prevents the muscles from forming, then it must be a closed hollow viscus into which the kidneys are secreting urine before the formation of those muscles takes place.

Now, according to Housden,[32] the urine is excreted solely into the allantois until the seventh week, and not until the ninth week does the urethra begin to carry the full urine load. If, as has been previously shown, the muscles are completely formed by the seventh week, at which time the urethra is only beginning to carry urine, then it is not possible for a urethral obstruction and bladder distention to prevent the ventral migration of the muscle tissue.

So we have now to consider the possibility of a secondary pressure atrophy of already existing abdominal muscles, a theory first advocated by Stumme[8] in 1903. He postulated that some form of urethral occlusion occurred late in embryonic life causing retention

of urine and a resultant hypertrophy and dilation of the urinary tract. Following the dilation of the bladder and ureters, there was either a pressure atrophy of the abdominal muscles or an embarrassment of circulation with ischemic atrophy. He believed that the obstruction disappeared later in development. This theory has been supported by Garrod and Davies,[9] Thatcher,[17] Housden,[32] Howard,[36] Dout and others,[38] Irvin and Krous,[39] and Eagle and Barrett.[42]

Facts that support Stumme[8] are: (a) The transversus, internal oblique, and lower half of the rectus are the muscles most frequently missing. These are followed by the external oblique and the upper rectus in that order. Thus, it is seen that the lower abdominal muscles are most often missing and they tend to disappear from within outward. (b) When fragments of muscle are present, they are almost always found at the costal and iliac margins, i.e., around the periphery of the anterior abdominal wall where pressure from a distended bladder would not be exerted. (c) The loose, thin, wrinkled abdominal wall visualized in all cases suggests previous massive distention. (d) In the cases reported by Pels-Leusden[12] and Ikeda and Stoesser,[21] there was no evidence macroscopically of abdominal muscle tissue, yet on microscopic cross-section, definite layers of fibrous tissue were found and interpreted by the pathologist as being the remains of muscle layers.

Arguments against Stumme's hypothesis are: (a) There are numerous cases on record of congenital urethral obstruction with dilated bladders and ureters, but completely normal abdominal musculature. (b) In the cases presented by Garrod and Davies[9] and

Lichtenstein,[35] there was no evidence microscopically of remains of muscle tissue.

3. There is a vascular defect which accounts for both the muscular and urogenital pathology. Dr. Bardeen, to whom Osler[7] referred his case, wrote that under normal conditions the abdominal musculature is preceded in the early covering of the abdominal wall by a vascular plexus, supplied from above by the internal mammary and from below by the epigastric artery. He postulated that an abnormal arrangement of blood vessels in the embryo prevented the formation of this plexus and impeded the growth of the abdominal musculature, and that, at the same time, circulatory disturbances gave rise to the abnormal conditions found in the bladder and ureters.

4. Deficiency of abdominal musculature may be caused by intra-uterine poliomyelitis. This theory has been disproved by three cases, Garrod and Davies,[9] Hall,[11] and Lichtenstein,[35] in which serial sections were taken through the spinal cord. The cells of the anterior horns were found to be normal in every respect.

Urogenital System.

1. Dilation and hypertrophy of the urinary tract may be secondary to an obstruction, structural or functional, at the urethra. This obstruction could be caused by any one of the following:

(a) Pull of the urachus or fibrous adhesion on the upper pole of the bladder creating a constriction of the vesicle neck.[8]

(b) Phimosis.[10]

(c) Neuro-muscular incoordination.[32]

(d) Prostatic hypertrophy.[32]

(e) Atresia.[36]

(f) Diverticula obstructing urethra when full.[37,42]

(g) Median bar fibrosis.[42]

(h) Valve-like folds in prostatic urethra.[40]

(i) A sharp folding or kinking of the penile urethra in utero could conceivably cause obstruction. This has not been proved in any case.

Arguments which favor this hypothesis are: (a) Those used to support Stumme's theory on pressure atrophy of the abdominal muscles. (b) The patent urachus reported as present in three cases, could be explained on the basis of lower urinary obstruction. (c) In the case of Daut and others,[38] the microscopic picture of the dilated ureters resembled those which become dilated later in life due to lower urinary obstruction. (d) Since Housden emphasized the importance of looking for an obstruction, it has been found in 40% of the cases. (e) Most of the cases have occurred in the male sex where urethral obstruction is most common.

Arguments against this theory are: (a) The same as those which conflict with pressure atrophy as the cause of abdominal muscle deficiency. (b) Most cases recorded have shown no urethral obstruction.

2. Lack of abdominal muscles (accessary muscles of micturition) causes excessive work, and therefore hypertrophy and dilation, of the urinary structures. Dr. Bardeen, referring to Osler's[7] case, wrote:

"It is possible that the lack of resistance, normally met with in the abdominal wall, may cause the bladder to expand rather than to empty the secretions into the amniotic cavity through the urethra." The bladder may then hypertrophy and while the fetus is still in utero, develop enough power to expell the urine through the urethra.

This theory is supported by the following facts: (a) The ones used to support a primary developmental defect of the abdominal musculature. (b) The urinary bladders in general have not shown the increased trabeculation that is usually present with organic urethral obstruction.

Objections to this hypothesis are those facts supporting urethral obstruction.

3. Attachment of the bladder to the anterior abdominal wall may cause excessive work and therefore hypertrophy of the bladder. This theory originated with Guthrie,[5] but has obtained little support since only eleven of the fifty-four cases show this attachment.

Gastro-Intestinal System.

Constipation has been a prominent symptom in the majority of cases and it has been noted that defecation is primarily an involuntary act. This condition is probably brought about by the deficiency of abdominal muscles which normally act as accessory muscles of defecation by contracting and increasing intra-abdominal pressure. When these muscles are absent the expelling force comes primarily from the smooth muscles in the wall of the rectum which are under the control of the autonomic nervous system.

An interesting feature of this syndrome which has stimulated very little discussion is the frequency of abnormal gut rotation. This anomaly has been encountered nine times in the thirty patients examined, and Silverman and Huany,[41] who were specifically looking for the defect, discovered it in all three of their patients. According to Dott,[43] thirty-five of his forty-eight collected cases of anomalies of the second stage of rotation, were accidental discoveries. He concluded that most cases must be asymptomatic, and the fact that seven of the nine cases considered here presented no symptoms of intestinal obstruction, substantiates his conclusion. Hence, we may theorize that, had all the patients in this series been examined carefully for minor malrotations, the incidence of gut abnormality would have been much higher.

Dott[43] explains this anomaly on the following basis: The loop of gut in the umbilical sac is withdrawn into the abdominal cavity at about the end of the ninth week. By this time the recti are already formed, and are normally constricting the opening of the sac, quickly closing it off when the gut is withdrawn. If the recti are deficient, it is possible that the orifice of the sac remains abnormally large and gut rotation is unable to take place in the usual manner.

Silverman and Huany[41] entertain the possibility that a generalized developmental arrest at about the sixth or seventh week accounts for the malrotation of the gut, the muscular and urogenital defects, and conceivably the other anomalies sometimes seen in this syndrome. In Hausden's[32] case the mother had contracted influenza

at precisely this time in gestation.

Thorax.

The thorax was described as deformed in twenty-five of the fifty-four cases. The abnormality was usually of the pigeon breast type with, at times, a Harrison's groove. Respiration has been observed as thoracic in many of the cases, while roentgenograms, when taken, have usually showed a flattened diaphragm.

With a deficiency of abdominal musculatures, this set of conditions might be anticipated. In abdominal breathing, the diaphragm contracts and thus flattens during inspiration. The tone of the abdominal muscles normally serves as the force which returns the diaphragm to the dome-like shape and accomplishes expiration. With a deficiency of the abdominal muscles, the diaphragm remains flattened and breathing must be accomplished by use of the thoracic musculature.

Some writers have speculated that this abnormal respiration, with its decreased vital capacity, has been a factor in the high incidence of pneumonia as a cause of death.(28)

Testes.

In one case,(42) the testes were found in the scrotum; the organs were not mentioned in twelve more. In every other patient cryptorchism was present and at every autopsy the glands were found in the abdominal cavity.

Housden(32) proposes that, since the testes descend in about the eighth month of gestation, the enormous bladder has already blocked the pathway to the inguinal ring, and has thus prevented their descent.

Those writers who believe that the primary defect in this syndrome is an agenesia of the muscle, explain the cryptorchism on the basis of abnormally low intra-abdominal pressure. This they feel prevents the proper formation of the processus vaginalis and the descent of the testes.(35) The scrotum, however, is practically always formed.

PROGNOSIS

The prognosis depends to a large degree on the amount of damage suffered by the urinary tract. As evidenced by Henderson's(3) and Levy's(13) patients, age sixty and seventy years respectively, deficiency of the abdominal musculature is compatible with long life providing the kidneys are in a good functional state. Neither of these patients had symptoms referable to the urinary tract.

However, since the incidence of kidney involvement has been high and death has generally occurred so early, the prognosis is definitely guarded until after a thorough investigation of the urinary tract has been made. With early institution of modern therapy the prognosis for life can be brightened considerably.(42)

TREATMENT

Problems in therapy include: (a) Correction of obstructive defects in the urinary tract. (b) Control of infections, particularly those of the urinary tract and lower respiratory passages. (c) Protection of abdominal viscera. (d) Management of

postural defects and overcoming difficulty in walking. (e) Alleviation of constipation.

In every case of defective abdominal musculature, the urinary tract should be explored early in life, even in the absence of symptoms. Pathology found is not consistent enough to warrant a definite outline of therapy, but separation of adhesions between bladder and umbilicus, resection of redundant ureters, resection or dilation of a urethral obstruction or any other procedure needed to establish a free flow of urine is indicated;[41] for it is only with this free flow of urine that further kidney damage can be prevented and that antibiotics are able to control urinary infections.

The use of abdominal support cannot be over-emphasized. Baxter's[28] patient learned to walk almost immediately when a corset was supplied. Defecation becomes easier and constipation is alleviated.[42] A well-fitted binder helps prevent the almost inevitable lordosis, and provides mechanical protection to the vital organs of the abdominal cavity.

CONCLUSIONS

1. Deficiency of the abdominal musculature is almost exclusively a disease of males and is accompanied usually by dilation of the urinary tract and cryptorchism.
2. The pathogenesis has not been definitely established.
3. Modern therapy offers those patients, with only moderate kidney damage, a good prognosis for a reasonably active life.

SUMMARY

A case of congenital deficiency of the abdominal musculature with associated urinary tract abnormalities is presented and the literature on fifty-three previous cases is reviewed.

The various theories on the pathogenesis are discussed. Most recent investigators subscribe to Stumme's[8] theory that the syndrome presented is initiated by urethral obstruction, either functional or structural, and that the dilation of the urinary tract, the deficiency of abdominal musculature, the pigeon breast deformity of the thorax and the cryptorchism are secondary manifestations following this obstruction in orderly and logical sequence.

The author tends to agree with this theory, but feels that, in view of: (1) Mollison's[14] case, where the muscle deficiency existed twelve years before the bladder was enlarged; (2) the cases of Lichtenstein[35] and Garrod and Davies,[9] where no remains of atrophied muscle was found at autopsy; and (3) the cases of Platt,[6] Smith,[34] and Silverman and Huany,[41] where the bladder was not apparently enlarged, there is sufficient evidence that the muscle deficiency is the primary factor to warrant further investigation of the subject. It is possible that one etiology will not explain all cases.

Modern therapy is outlined and the prognosis discussed.

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