Craniospinal Malformation in Four Siblings

Case Report

LEONARD A. TITRUD, M.D.*
Minneapolis, Minnesota

When one child is born with a congenital anomaly there is an appreciable tendency for siblings to be malformed also. The abnormalities involving the children of the family described here occurred in the first 4 offspring (3 boys and 1 girl); whereas 3 children born later (a 3- and 7-year-old daughter and a 5-year-old son) are normal. The parents have no congenital malformation nor is there any among known relatives.

A ten-year study of meningoceles and meningomyeloceles in Atlanta in 1952-1961 revealed the incidence of 76 per 100,000 white live births and 28 per 100,000 nonwhite live births! Murphy found that for every 218 infants who were born alive, 1 was malformed at birth, and that families having a deformed child subsequently had an abnormally developed infant at a rate of frequency about 25 times greater than that for families with normal children. In families possessing 2 or more malformed offspring, the congenital defect that involved the first infant will be duplicated in about one-half of siblings born later. Murphy also learned in his study that the duplication of defects occurred in the mother’s relatives almost 3 times more frequently than in the father’s.

Approximately 60 per cent of all the developmental defects of newborns involve the central nervous system, hydrocephalus with or without spina bifida being the most frequent. Ingham and Lowrey found that 25 per cent of normal children have occult defects in the vertebral laminae, these occurring most commonly in the lumbar and sacral regions. Campbell indicated that during development of the human embryo, spina bifida or cranium bifida with or without meningocele or encephalocele seems to occur at about the 21st to 29th day with a failure of closure of neural tube. Bentley and Smith emphasized the occasional even more complete lack of fusion of the blastopore with the resultant syndrome of split notochord so that intestinal fistulae may actually extend posteriorly between the divided structures of the vertebral column, and that other defects such as diastematomyelia, diplomyelia and spina bifida may be explained on this basis also.

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* Address: 929 Medical Arts Bldg., Minneapolis, Minnesota 55402.

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Case 1. M.F., a white male, was born Jan. 29, 1949, the first of 7 children of the same mother. The birth was normal. Physical examination showed an upper lumbar meningocele, 4 cm. in diameter. There was also a small occipital pilonidal-sinus dimple. The head was normal in size. Neurological findings were normal, as were roentgenograms of the skull and chest on March 1, 1949.

On March 5, 1949, with the aid of local anesthesia, the sac of a lumbar meningocele, 5×6 cm. in diameter, was excised and the defect over the area of the spine bifida at the lumboconal juncture was closed. The sac of the meningocele was multilocular and did not contain any elements of nerve.

Examination on Jan. 18, 1956 showed that this boy was normal. His head seemed somewhat larger in proportion to his body. On March 16, 1964 he was well and able, except for a recent injury to his back.

Case 2. S.F., a male and the second offspring, was born March 14, 1951. He weighed 7 lbs. and 8.75 oz. at birth. The pregnancy and delivery were normal. Findings were normal except for a thoracic meningocele, 4 cm. in diameter. On March 19, 1951, roentgenograms of the thoracic spine showed defects of spine bifida from the 3rd to 5th thoracic levels. There was no hydrocephalus.

On May 10, 1951, with general anesthesia, the meningocele was excised and the defect was repaired. There were no elements of nerve in the sac.

This boy was entirely well and normal on examinations, Jan. 13, 1956 and March 16, 1964.

Case 3. L.R.F., the third child, was a white male born on July 7, 1954 after a 40-week pregnancy and labor of 2 hrs. and 17 min. The weight at birth was 7 lbs. and 7.25 oz. This child had a thin-walled suboccipital meningocele, 7 cm. in diameter. There was also an upper thoracic meningocele, 4.5 cm. in diameter. The cranial sutures were wide and the circumference of the head was 32.5 cm. on July 7, 1954. By July 26, 1954, the cranial circumference was 35.5 cm. and there was a prominent distention of the anterior fontanel. The suboccipital and thoracic meningoceles were distended and tense. Roentgenograms of the skull showed moderate Luckenschädel, and the large occipital soft-tissue mass was interpreted to be an encephalocele. A similar but smaller mass at the 9th thoracic vertebra was that of a meningocele. The spine at the level of the 1st lumbar vertebra was typical of diastematomyelia.

Operation was performed on July 27, 1954 with the use of local anesthesia. A venous cutdown in the ankle was used for the administration of fluids and blood. The suboccipital encephalocele-meningocele, which contained 150 cc. of dark colored fluid, was excised. This cyst seemed to arise at the level of the 1st and 2nd
cervical vertebrae. The child died from respiratory failure during the operation.

Autopsy showed a cranial defect, 1 by 1.5 cm., adjacent to the confluent sinus. The spinal cord showed hydromyelia through the cervical and thoracic portions. In the midlumbar area there was a bony spur typical of diastematomyelia extending through the spinal cord.

Case 4. J.J.F., was a white girl born Jan. 10, 1956, after a 41-week pregnancy and normal labor. The weight at birth of this fourth child was 8 lbs. and 14 oz. The head appeared small and the cranial sutures were widely separated. Roentgenograms of the skull on Jan. 13, 1956 showed a large foramen magnum and an occipital encephalocele, 8 cm. in diameter. The baby was rather quiet and listless. Physical and neurological findings otherwise were normal. There was no increased intracranial pressure.

On Jan. 20, 1956, with the use of 10 gr. intrarectal chloral hydrate and local anesthesia, the sac of the encephalocele which communicated with the subarachnoid space was excised. The baby continued to be listless with some reduced blood pressure and depth of respiration and exhibited some twitching movements of the left extremities. She died on Jan. 23, 1956.

Autopsy showed a wide separation of bone at the anterior fontanel. There was a prominent deficiency in the formation of the tentorium and a marked agenesis of the cerebellum. The 3rd ventricle was abnormally wide. The pons and medulla oblongata were not distinctly recognizable; there were some small globular elements of brain tissue adjacent to the area of the cranial defect.

Summary and Conclusion

The rare involvement of so many siblings with cranial and spinal congenital defects emphasizes the possibility that heredity may be a factor in the transmission of such malformations.

References