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DEVELOPMENTAL DISORDERS

PROGNOSIS OF OPEN SPINA BIFIDA

The outcome of 117 consecutive cases of open spina bifida treated unselectively from birth between 1963 and 1970 and reviewed 16-20 years later is reported from the Department of Urology, Addenbrooke's Hospital, Cambridge, England. Forty-one percent died before their 16th birthday and most of the survivors were badly handicapped: 50% were wheelchair dependent, 30% were mentally retarded and 75% relied on an appliance or padding for incontinence. The majority had had a shunt inserted within hours of birth. 75% were incapable of employment and 50% were unable to live without help or supervision. The sensory level at birth provided a yardstick for predicting the range of handicap into adult life and was useful in counseling parents concerning the child's long-term prognosis. With a sensory level above T11 the disability would be severe, with no prospect of walking or of continence and with a 10% chance of independent living. A sensory level between T11 and L3 (from the umbilicus to the knees) should indicate a moderate disability in 60% and a capability of living independently in 45%. A sensory level below L3 should predict a survival rate of 75% and a normal intelligence in 80%; 90% would be ambulant as adults and 45% would be continent; and 85% would be totally independent with moderate or minimal disability. (Hunt GM. Open spina bifida: Outcome for a complete cohort treated unselectively and followed into adulthood. *Dev Med Child Neurol* Feb 1990; 32:108-118).

COMMENT. The impact of treatment on disability is far less than that on mortality. In groups of patients treated selectively, the mortality among the untreated cases is higher and those selected for treatment are born with a much lesser

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degree of disability. Thoracic scoliosis and horseshoe kidney that may be recognized during fetal life are associated with sensory levels in the thoracic region and are predictive of a poor prognosis. The author points out that the reliable discrimination between mild and severe cases of spina bifida in early pregnancy is not yet possible and parents should be informed of the likelihood of prolonged dependency into adulthood.

ANENCEPHALY

The Medical Task Force on anencephaly coordinated by Stumpf DA (Northwestern University Medical School, Chicago) presents a consensus statement limited to medical issues of organizations of physicians caring for fetuses and infants with anencephaly. The statement was approved by the AAP, AAN, ACOG, ANA, and CNS. Anencephaly is defined as a congenital absence of a major portion of the brain, skull, and scalp with its genesis in the first month of gestation. The primary abnormality is failure of cranial neurulation, the embryologic process that separates the precursors of the forebrain from amniotic fluid. Anencephaly does not mean the complete absence of the head or brain. Craniofacial anomalies are associated and up to 1/3 have defects of the non-neural organs that could preclude their use for transplantation. The maternal serum a-fetoprotein level is elevated in 90% of cases, and elevated a-fetoprotein levels in amniotic fluid and the presence of acetylcholinesterase on electrophoresis occur in virtually all cases. Ultrasonography is also reliable in the prenatal diagnosis of anencephaly. The postnatal diagnosis requires the following criteria: 1) Absence of a large portion of the skull, 2) absence of scalp over skull defect, 3) exposed hemorrhagic fibrotic tissue, 4) absence of recognizable cerebral hemispheres. The cause is usually not known and a polygenic or multifactorial etiology is suggested. Chromosome abnormalities and mechanical factors are recognized associations. Hyperthermia and deficiencies of folate, zinc and copper in the mother have been invoked. In recent years, 80-90% are aborted, 7-10% are stillborn, and 3-11% are live born. Most live born anencephalic infants have died within the first days after birth and survival beyond one week occurred in 0-9% in three series. Two months was the longest survival confirmed with accepted diagnostic criteria. The estimated incidence of anencephaly in the U.S. is 0.3-7/1000 births and the incidence of live born infants with anencephaly would be less than 100 per year. Anencephalic infants have no functioning cerebral cortex and are permanently unconscious. Brain stem functions are present in varying degrees and the diagnosis of brain stem death depends on the disappearance of previously existing brain stem functions, including loss over an observation period of at least 48 hours of measurable cranial nerve function and spontaneous movements, and a positive apnea test. Confounding factors such as drugs, hypothermia, or hypotension should be excluded. The use of organs from infants with anencephaly for transplantation is also discussed. (Stumpf DA et al. The infant with anencephaly. N Engl J Med March 8, 1990; 322:669-674).