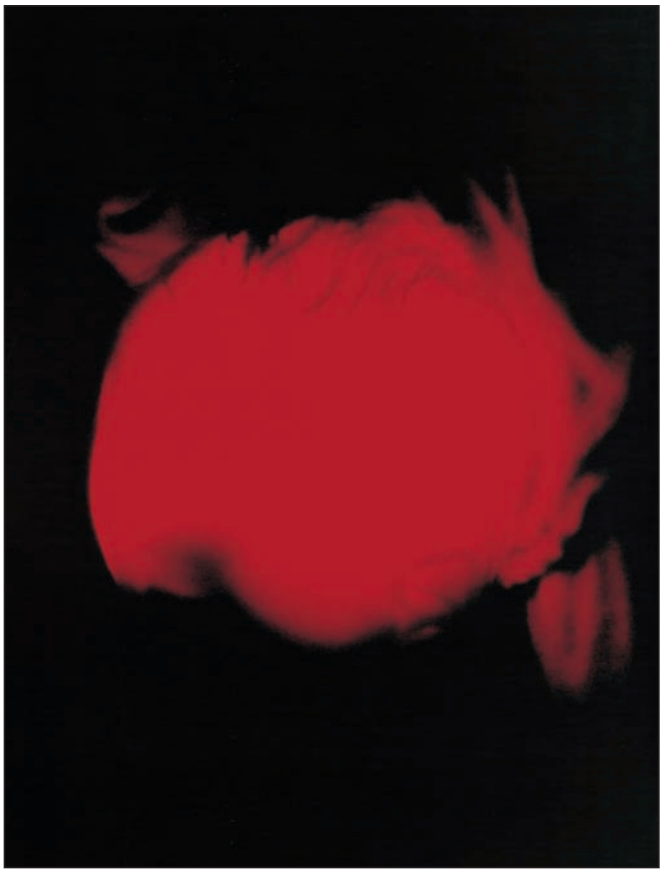
Transillumination of the neonatal skull: seeing the light

An infant was delivered vaginally at term to a healthy 21-year-old primiparous woman in a community hospital. There were no immediate neonatal concerns. There had been no antenatal exposure to drugs or alcohol, or report of flu-like illnesses or rashes, and the pregnancy was unremarkable except that the mother did not have an 18-week ultrasound for fetal anatomy. On day 1 of life, decreased tone and poor feeding were noted, and the baby was transferred to a tertiary care centre.

Findings on general physical examination were unremarkable. The infant's weight was 3.6 kg and the head circumference was 34 cm (normally 32.5–37.0 cm). The head and neck were normal on gross examination, and the anterior fontanelle was noted to be full but not bulging. On neurologic examination the baby had normal deep tendon reflexes but generally decreased muscle tone. The Moro and sucking reflexes were also depressed. Transillumination of the skull was performed using a bright light source (in this case a Welch Allyn Lite Pipe Neonatal Transilluminator) applied to the anterior fontanelle. Fig. 1 demonstrates a face-on view with transillumination of virtually the entire skull and even the right pupil. Fig. 2 also demonstrates significant transillumination of the skull, but from a posterolateral view. An ultrasound of the head and a subsequent CT scan revealed absence of the cerebral hemispheres (hydranencephaly) and abnormal development of the brainstem and cerebellum. The infant suffered aspiration and respiratory failure and ultimately died at 2 weeks of age.

Hydranencephaly, a condition in which most or all of the cerebral hemispheres are absent and are replaced by cerebral spinal fluid, has been reported in 0.2% of infant autopsies.[1] About 1% of infants thought to have hydrocephalus clinically are later found to have hydranencephaly. The condition can usually be differentiated from severe hydrocephalus (porencephaly) by neuroimaging or post-mortem examination. The cause is not clearly understood, but hydranencephaly is felt to occur secondary to a destructive phenomenon such as vascular occlusion or infection. The prognosis is extremely poor, with severe neurologic dysfunction, seizures, myoclonus, respiratory failure and death usually occurring within the first few days to weeks, although rare reports of prolonged survival have been documented.[2]

This case illustrates the usefulness of transillumination of the skull, also known as skull diaphanoscopy, as a



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clinical tool in the evaluation of neonates and infants with suspected neurologic disease. Transillumination of the skull was first described in 1831 by Richard Bright[3] and was later recognized as the first form of light-based diagnosis of hydrocephalus. Over time, the technique was modified and used to diagnose intracranial hemorrhage in the neonate before the availability of ultrasonography.[4] It has since been used as a screening procedure for infants with macrocephaly and those suspected of having a subdural effusion, subdural hematoma, hydrocephalus, hydranencephaly, porencephaly, increased intracranial pressure and even skull fractures and nutritional deficiencies.[5–8]

Attempts have been made to develop standardized light sources[9] and measurement standards based on age,[10] but these are not in wide use. In general, when using a standard 2-cell flashlight held tight to the anterior fontanelle, transillumination of more than 2 cm around the edge of the beam or asymmetry of the transillumination suggests underlying pathology. However, this parameter will vary significantly depending on prematurity, age, light source and operator technique. Given its low cost and simplicity to apply, transillumination is a useful initial screening test for infants with abnormal findings in the office or in hospital settings where CT or MRI scanning may not be immediately available. With advancements in and increased availability of neuroimaging techniques, transillumination has become a somewhat forgotten tool, but sometimes even a simple otoscope[11] placed in the appropriate position can provide clinical "enlightenment."

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