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Case report

Prolonged survival to adulthood of an individual with hydranencephaly

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Abstract

Hydranencephaly is a rare condition in which the brain's cerebral hemispheres are absent. The outlook for children with hydranencephaly is generally poor, and many children with this disorder die during infancy. Although rare, patients with prolonged survival have been reported. A 22-year-old female patient was admitted to our hospital in a bedridden state for management of bedsores. The initial brain computed tomography showed complete replacement of the cerebral hemispheres with fluid and no cortical plate or hemispheric white matter. The bilateral brain stem, some of the left cerebellum, and the inferior frontal lobe were spared. Although certain behaviors were observed, such as eye opening in response to sound and painful stimuli, examination identified neither a definite awareness of the environment nor a meaningful action. To our knowledge, this is the longest survival of a patient with hydranencephaly in the world.

Keywords: Hydranencephaly; Survival; Prolonged; Adult

1. Introduction

Hydranencephaly is characterized by a marked reduction in total brain substance, which is replaced largely by fluid within a relatively normal-size cranium [1]. Patients with hydranencephaly typically have a markedly reduced life expectancy, either being stillborn or dying within a few weeks or months after birth [2,3]. Exceptionally prolonged survival has been reported rarely, but specific neuroimaging or neuropathological details to confirm the diagnosis or ascertain the putative pathophysiological features to account for the prolonged survival are often lacking. Here, we describe the clinical and radiological features of an adult patient with hydranencephaly who survived to 22 years and 6 months of age. To our knowledge, this is the longest survival of patient with hydranencephaly in the world.

2. Case report

The patient was 22 years and 6 months old. Her parents were not known and had abandoned her as a newborn, after which she was cared for by public pediatric hospitals or nursing homes. Recently, she was delivered to our hospital to receive care for a bedsore and referred to our neurology department for general assessment. Her past history showed various infections such as pneumonia. However, her general condition at the initial visit to our department was relatively good. She was in a bedridden state and was maintained with a nasogastric tube and Foley catheter. Her head circumference was 70.5 cm and her facial appearance showed no anomalous features (Fig. 1). Her body was underdeveloped for her age and showed generalized spastic muscle tone with deformities of all extremities. Although certain behaviors were observed, such as eye opening in response to sound and painful stimuli, examination demonstrated neither a definite awareness of the environment nor meaningful actions. The pupillary response to light was minimal. Visual response to threat was not elicited. The eyeball did not move spontaneously and did

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Fig. 1. General appearance of the patient. She shows completely flaccid muscle tone throughout the body, including the limbs and neck, but no facial anomalies.

not responds to objects moving around her. Gag reflex, sucking, grasp reflex, other primitive reflexes, Babinski's sign, and other pathological reflexes were not observed.

Brain computed tomography (CT) showed complete replacement of the cerebral hemispheres with fluid and no visible cortical plate or hemispheric white matter. The bilateral brain stem, some of the left cerebellum, and the inferior frontal lobe were spared (Fig. 2). Electroencephalogram revealed complete isoelectric potential, the so-called "electrocerebral silence" from all recorded sites according to the International 10–20 System.

The patient was diagnosed with adult hydranencephaly. After checking all the records about her and her initial registration as a baby, we were able to identify that her age was at least 22 years and 6 months according to the initial registry about her birth. However, we could find no records about her parents or family, or the perinatal condition of her birth.

3. Discussion

Hydranencephaly is a term of gross description that includes a group of disorders that vary widely in their presumable time of onset, mechanisms of pathogenesis, regions of involvement of the central nervous system (CNS), and anatomic organization of the brain remnant [1]. Although remnants of a nonfunctioning cortex can be present, the hallmark is the extensive reduction in brain matter that has been replaced with cerebrospinal fluid [1]. Facial features are uniformly normal, and these distinguish hydranencephaly from other major CNS anomalies [4]. Typically, hydranencephaly arises prenatally and is related to either a developmental or an encephaloclastic process, often of infectious, toxic, or genetic origin, that affects the major vessels of the anterior circulation [4–6]. Postnatal insults can also cause a similar pathological and neuroimaging appearance [7].

A previous report noted that survival of the hydranencephalic infant for more than a few weeks requires integrity of the hypothalamus, as indicated by the ability to regulate body temperature [1]. However, abnormal hypothalamic–pituitary function has occurred in prolonged survivors [4,8]. Prolonged survival might also be expected to correlate with the absence of associated malformations in other organs. Nevertheless, most previous reports did not include extensive evaluation for non-CNS malformations among long-term survivors. In addition, both normal and abnormal neurophysiological tests can occur in prolonged survivors. For example, the presence of electroencephalographic activity is not needed for prolonged survival because many patients exhibit an isoelectric or near-isoelectric electroencephalographic recording. Similarly, absent visual-evoked and brainstem-evoked, and abnormal somatosensory-evoked potentials are evident in prolonged survivors [4].

McAbee et al. recently published a survival analysis of patients with hydranencephaly [4], which demonstrated that prolonged survival up to 19 years can occur in individuals with hydranencephaly, even those lacking rostral brain regions, who show isoelectric electroencephalograms. However, this review failed to suggest a definitive set of features that are predictive of prolonged survival and did not indicate a clinical or neurophysiological predictor of prolonged survival with hydranencephaly.

Our patient appears to be the longest surviving individual with hydranencephaly; she has lived for about 3 years and 6 months longer than the previously reported longest surviving patient. She is still alive and in a relatively healthy state, and her expected lifespan cannot be determined. This case shows that a patient with hydranencephaly can live beyond adolescence to adulthood. We also examined her carefully for any distinguishable clinical or neurophysiological features to distinguish her from other shorter-surviving patients, but we found no features to predict her long survival. Similar to previous patients, her brain showed complete replacement of the cerebral hemispheres with fluid, and no cortical plate or hemispheric white matter were visible. Structures of the posterior fossa such as the bilateral brain stem and some of the left cerebellum were spared. We also identified electrocerebral silence from her electroencephalogram. Hence, we believe that the clinical and neurophysiological predictors for long-term survival in hydranencephaly cannot yet be determined based on our case report and previous case reports.

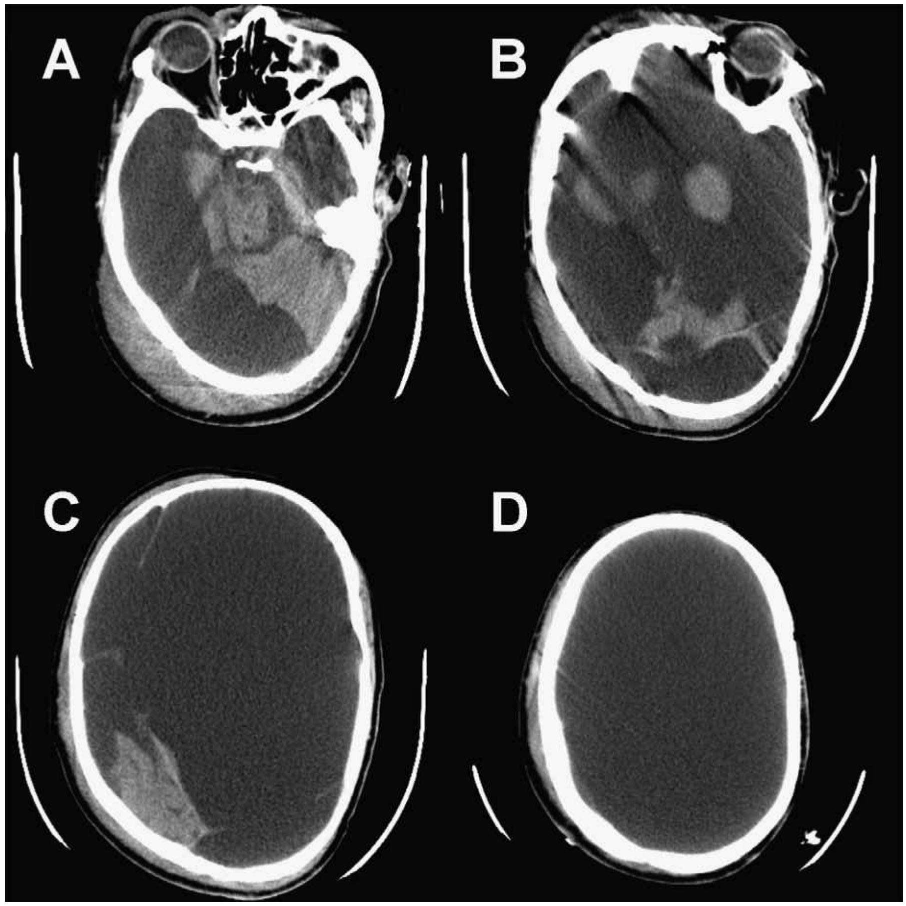

Fig. 2. Computed tomography (CT) features of the patient. Brain CT shows complete replacement of the cerebral hemispheres with fluid and no visible cortical plate or hemispheric white matter (D). The bilateral brain stems, some of the left cerebellum (A), presumably some of the caudate head (B), and a remnant of the occipital lobe (C) are spared.

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