A case of complete absence of the visual system in an adult. By William G. Spiller, MD, Assistant Clinical Professor of Nervous Diseases and Assistant Professor of Neuropathology in the University of Pennsylvania; Clinical Professor of Nervous Diseases in the Women’s Medical College of Pennsylvania. [From the William Pepper Laboratory of Clinical Medicine (Phoebe A Hearst Foundation)] Brain 1901: 24; 631–642.

Dr Spiller argues that in studying the complete absence of the visual system in an adult, the potential confounding effect of imperfect development affecting other parts of the nervous system present in ‘agenesia of the visual system in a newborn child’ are avoided. Born in 1878, T.S. is admitted to the Pennsylvania Training School for Feeble-minded Children in March 1895. ‘He was an idiot, absolutely helpless, and very unclean…[but] he was passionately fond of music’. Dr Spiller’s initial diagnosis is cerebral spastic paraplegia of the lower limbs, with absence of the eyeballs. T.S. weighs 38 pounds and is 3 ft 7 in. tall. Unable to support his own weight, T.S. can be helped to take a few ‘scissored’ steps. He mutters monosyllables and can only say ‘mamma’. The palpebral fissures are small. Despite being aged 22 years, T.S. is prepubertal and has the physical appearance of a child. He remains institutionalized for the rest of his life, dying on 2 March 1900. Autopsy is performed 2 days later (Fig. 1).

There are flexion contractures of the arms and legs. Each orbit contains a small amount of fibrous connective tissue but nothing resembling an eyelid is present. There are no optic foramina, merely imperforate depressions in the skull at the expected place. No trace of the optic nerves, chiasma or tracts can be found. Nor is there any sign of an external geniculate body on either side (Figs 2 and 3). The brain is small and firm with selective atrophy of the left ascending frontal convolution. The occipital lobes are also small with a short calcarine fissure. These are the preliminary observations and Dr Spiller now fixes the brain for subsequent sectioning and staining with thionin and Weigert’s haematoxylin.

There is a slight reduction in the number of nerve cell bodies from the third, fourth and fifth layers of the (left) calcarine fissure with depletion of giant Betz cells from layer three, and of the projecting fibres of Vicq d’Azyr. Moving forward, the optic radiation is present but small. Meynert’s commissure is judged normal; however, no trace can be found of the external (lateral) geniculate body. Referring to the anatomy depicted in Jules and Augusta Dejerine’s Anatomie des Centres Nerveux (1895–1901), the optic tracts cannot be found. Nor is there anything much resembling the expected density of nerve fibres within the pulvinar and posterior thalamus. In contrast, although cell bodies of the oculomotor and trochlear nerves are normal, those of the abducens nerve are reduced and the nerve fibres small, demyelinated and ‘sclerotic’. The density and appearance of anterior horn cells in the spinal cord are normal.

But does documenting tissue that is missing alongside parts that are present in an adult with anophthalmia resolve the debate on which structures are essential anatomical components of the visual system? Constantin von Monakow has argued that the primary optic centres are the external geniculate body (receiving 80% of fibres from the anterior visual pathway), the surface of the pulvinar and the superficial grey matter of the anterior colliculi of the corpora quadrigemina. In addition, Dr Spiller acknowledges the views of Benedikt Stilling, Bernheimer and Albert von Kolliker that there may be a contribution from the subthalamic nuclei, the internal geniculate body and the tuber cinereum. Overall, his examination of T.S. supports the conclusions of von Monakow, but also confirms that Meynert’s commissure and the

Figure 1 A photograph of T.S. taken after death. The right upper limb, the fingers of both hands and the lower limbs are slightly contractured. The palpebral fissures are small.
habenula are not part of the visual system. von Monakow had also considered that centrifugal fibres, originating in the giant pyramidal neurons of layer three in the visual cortex, pass in the optic radiations to the anterior colliculus of the corpora quadrigemina. Dr [nk] von Leonowa has studied four neonatal cases of anophthalmia and supports the view that there is an intimate relationship between vision and development of the anterior colliculi of the corpora quadrigemina and the external geniculate body but not the subthalamic nucleus. She has observed that the nuclei of the third, fourth and sixth cranial nerves are intact in cases of anophthalmia if the ocular muscles are present within the orbit. But Dr Spiller finds normal oculomotor and trochlear, yet not abducens, nerves; apparently in the absence of any orbital structures (although there may have been muscle inside the small amount of fibrous tissue within T.S.’s vestigial orbit). How can this be?

Again, von Leonowa has described a foetus in which—in the absence of a brain or spinal cord—the vertebral canal was filled with non-medullated nerve fibres, or with a very thin myelin sheath, originating from dorsal root ganglia and projecting centripetally to end in...nothing. ‘The muscles of the limbs appeared fully normal, although no motor nerves existed.’ Others have reported broadly similar findings of central projections from dorsal root ganglia wandering into a spinal vacuum. Dr Spiller accepts that: ‘in early life muscles may be developed, although no motor nerves exist...[and]...the converse may well be true – i.e. motor nerves probably may exist, although the muscles are not developed’.

As for Meynert’s fibres, believed to connect the optic nerves and the oculomotor centres, some are now considered also to project to the anterior columns of the spinal cord. Dr Spiller finds these intact in the case of T.S., indicating that their role is not just to subserve the pupillary reflexes (Fig. 4). The lack of neuronal depletion in the layers of the calcarine sulcus in cases of anophthalmia is not so surprising. These nerve cells project to the mid-brain visual centres already described and are not directly concerned with vision. Rather, they form part of complex association networks that integrate with systems primarily concerned with movement of the eyes and limbs, and with speech (Fig. 5). ‘It is not surprising that these association neurones are developed even in a case of congenital defect of the visual system...[but]...it is difficult to understand what the function of these fibres [in the case of T.S.] might be.’

As for T.S.’s idiocy, clearly this cannot be attributed solely to the lack of a visual system. Despite having a spastic paraplegia in life, the anterior horn cells are not depleted and there is no sclerosis of the lateral columns: ‘this is not the first time I have found an

Figure 2 The occipital lobes have been cut away. A portion of the right temporal lobe has also been cut away in order to show the complete absence of the visual system. The internal geniculate body (A) is well shown, but no trace of the external geniculate body or of an optic tract or chiasm can be found.

Figure 3 The optic thalami are well shown. The anterior colliculi of the corpora quadrigemina, especially the right colliculus, appear smaller than the posterior, because the former were further from the camera.
apparently normal spinal cord in congenital spastic paraplegia, and I think the explanation is to be found in an imperfect development of the motor neurones... sclerosis may be entirely absent if destruction of nerve fibres occurs in early life, because the growing tissue of the spinal cord fills the space left by degenerated fibres, and proliferation of the neuroglia does not occur'. T.S. is not unique; another inmate of the Pennsylvania Training School for Feeble-minded Children had been affected by idiocy and partial visual loss attributed, at autopsy, to absence of the optic chiasm. Additionally, Otto Haab has described a 27-year-old woman with the mind of a young child, in whom normal oculomotor nerves and muscles were present in the context of only vestigial traces of the optic tracts, with complete absence of the external geniculate body and reduction in size of the pulvinar of the optic thalamus.

Taken together, Dr Spiller feels able to conclude that the primary optic centres are the external geniculate body and the thalamic pulvinar; the anterior colliculus of the quadrigeminal body is relatively unimportant for vision; and the subthalamic nucleus, habenula and internal geniculate body are not involved. The cellular architecture of the calcarine fissure and adjacent sulci are unaffected by the lack of an anterior visual pathway and eyes; the oculomotor nerves and muscles develop independently of the visual system; and congenital spastic paraplegia is not necessarily marked by abnormalities of the spinal cord.

William Spiller’s description of the preservation of anatomical structures within a single sensory system despite the complete absence of that ‘sensation’—in this context vision—is taken up in the current issue by Holly Bridge and colleagues from Oxford, who show predominantly subcortical abnormalities, and much preservation of structure and connectivity of the visual cortex and its relationships to the external (lateral) geniculate body, in individuals with congenital anophthalmia studied in life by magnetic resonance imaging (page 3467).

Alastair Compston
Cambridge