Closure of the eyelids—voluntarily or as part of the reflex response to corneal stimulation, noise or visual threat—involves coordinated contraction of the orbital and preseptal portions of the orbicularis oculi muscles with relaxation of the elevator of the upper lid. Perhaps the pretarsal portion of the muscle is preferentially activated in the reflex movements and the more peripheral parts in voluntary eye closure. Ralph Ross Russell describes three patients, each with Creutzfeldt–Jakob disease, having a variant of supranuclear palsy in which voluntary and reflex eyelid closure are dissociated.

A professional musician, aged 52 years, complains of difficulty using rapid movements of the left hand and he cannot close his eyes unless by gently pressing downwards on the upper lids with his fingertips whereupon they remain shut but can voluntarily be opened. His eyes close spontaneously during sleep. Neurological examination confirms the dissociation between impaired voluntary eye closure and intact reflex blinking, and (initially) in the absence of other signs apart from a brisk jaw jerk. Over the next 6 months, he progresses through stages of mild upper motor involvement of the bulbar and limb musculature to impairment of voluntary and pursuit eye movements and a complete inability to close the eyes, although the lids continue to move during down gaze. The disorders of bulbar function, eye movement and limb spasticity increase and are eventually associated with focal muscle wasting and fasciculation, and dementia leading to death at 2 years after presentation.

A motor mechanic, aged 32 years, complains of difficulty using the right hand and slurred speech. He has a pseudobulbar palsy and upper motor neuron signs in the limbs. After 12 months slow progression of these complaints, and the emergence of dementia, he develops difficulty with ocular refixation such that he can only alter his direction of gaze by closing the lids, moving his head and opening on the newly fixated point—this limitation occurring in the presence of preserved but slowed pursuit movements and intact optokinetic nystagmus. As his bulbar and limb spasticity increase over the next few months, and at a time when ocular movements are retained but slow, he loses the ability to close his eyes voluntarily but has preserved reflex movements of the eyelids. Bell’s phenomenon is lost. Remaining in this parlous state for some time, he dies about 4 years after onset.

A marketing-consultant, aged 35 years, develops mild dysarthria that worsens over months and is then associated with altered mood; a year later he has developed a supranuclear gaze palsy, predominantly in the horizontal plane, and—although able to close his eyes voluntarily—he cannot blink; during the final illness he has dementia, pseudobulbar palsy, inability to close the eyes to command but preserved reflex movements of the lids, loss of Bell’s phenomenon, quadriplegia and myoclonus. Death occurs about 2 years after onset (Fig. 1).

All three patients have Creutzfeldt–Jakob disease confirmed at autopsy and manifesting widespread cortical neuronal loss, pyramidal tract degeneration and loss of myelin, marked status spongiosus especially affecting layers 2 and 3 of the cortex, and superficial cortical gliosis. Investigations available in life had not proved useful and the patients were thought to have a variant form of motor neurone disease; in no case was the diagnosis of Creutzfeldt–Jakob disease suspected during life. One purpose of the paper is to draw attention to progressive symmetrical supranuclear facial and oculomotor paresis as early features of the disease and this has since been confirmed in other reports. Loss of rapid voluntary blinking is followed by impaired sustained eyelid closure except with manual assistance. Opening the eyes is unaffected and reflex eye blinking occurs in response to the usual stimuli. Previous examples of this syndrome are reported in cerebrovascular disease—although most patients with bilateral ischaemic lesions, facial diplegia and pseudobulbar palsy are able to close their eyes voluntarily—and in motor neurone disease. Despite extensive analysis, and theoretical considerations relating to lesions of the dominant supramarginal gyrus, voluntary loss of eyelid closure is not described as part of the facial apraxias. It seems logical to regard the defect of eye closure as a variety of supranuclear palsy caused by bilateral loss of cortical motor neurones in the facial areas of the motor cortex. This explanation is consistent with the associated loss of Bell’s phenomenon, also reflecting altered fronto-pontine connections, whereas preservation of reflex blinking in response to noise and visual threat confirms intact brainstem and occipito-pontine pathways. As for the other abnormality of eye movement observed in these patients, difficulty in transferring gaze in the horizontal plane, Ralph Ross Russell has as his authority an earlier description by Sir William Gowers from 1879.
Keeping the eyes fixed on a point of interest depends, according to Dr Gowers, not only on the influence of a voluntary centre but also on reflex factors in which visual input from the retinal ‘fixation’ point feeds forward to the motor impulses. But Gowers is not aware of any physiological or pathological evidence that does in fact demonstrate such reflex activity. In the agonal stage of progressive muscular atrophy with rigidity, a patient at the National Hospital for the Paralysed and Epileptic is observed to have great difficulty in moving his eyes between two objects displaced by 45°. He achieves this by first turning his head and then allowing the eyes gradually to follow and align in the mid-position on the new object of interest. Of course, the first step in this manoeuvre must involve a rapid movement of the eyes equal and opposite to that of the head initially leaving them eccentrically fixed on the first object. This is against the ‘will’ and so must be driven by a powerful visual reflex urge. In this case, the usual subordination to the will has escaped, revealing an independent existence of the reflex stimulus. But that there should be an effortless means of keeping the eyes fixed during movements of the head, which develops and is strengthened during infancy by increasing awareness of the rich visual environment, does make sense. The seat of this reflex process is probably the corpora quadrigemina. By analogy, this reflex inertia of the eyeballs is not dissimilar to the rigidity and slowness of limb movements seen in the same patient which can also be interpreted as reflex, the stimulus coming from the muscles themselves and now subject to reflex responses dissociated from the normally over-riding influence of the voluntary centres, or at least indicating a resistance to the action of the latter that is only slowly overcome.

Ralph Ross Russell takes up Gowers’s prescient presherringtonian analysis of voluntary eye movements, concluding that—together with the absent Bell’s phenomenon and supranuclear palsy of eyelid closure—loss of voluntary ocular refixation is also due to bilateral lesions of the fronto-pontine pathways.

Fig. 1 Case 3—eyelid closure (6 months before death). (A) Attempted voluntary eye closure on command. (B) Blinking to visual threat. (C) Blinking to glabellar tap. (D) Attempting to close eyelids digitally. (E) A random blink. (From RW Ross Russell).
starting with slow, irregular and ill-sustained horizontal voluntary gaze whilst normal pursuit and reflex movements are still preserved. Eventually, the normally latent occipital fixation and pursuit reflexes come to dominate the eye movements and there is spasm of fixation. This reminds Dr Ross Russell of Sir Gordon Holmes’s dictum that voluntary rapid eye movements should be tested in two ways, first in the absence of fixation (in front a blank wall) and second in a normal visual environment with objects of visual interest.

Twenty-nine years on, Ralph Ross Russell recalls that this paper was first written as a shorter report illustrating the dissociation of voluntary and reflex facial movements in supranuclear palsy, but publication was delayed because of editorial reservations at Brain over the propriety of including the photographic illustrations. As submitted, these comprised a number of frontal full-face views of the face and eyes and it was not possible adequately to disguise the patients’ identities. Case 1 was a well-known concert pianist and publication was considered by the Deputy Editor to be an ethically unacceptable breach of professional confidentiality. Unhappily, the early death of the patient, the discovery of the underlying pathology and the permission of the relatives were sufficient to calm these editorial scruples and publication was eventually agreed.

As the paper by Matteo Bologna and colleagues from Rome (Italy) in the current issue shows (page 502), the study of eye movements continues to inform our understanding of organization and disorganization in the CNS, providing a window on the complex integration of pathways within the brain stem and their modulation by connections from many parts of the cerebral cortex, that illuminates the nature of these and all other voluntary and reflex movements orchestrated by the CNS.

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