Axial T1 weighted MRI 48 hours after surgery. Complete tumour removal was documented in all planes. The section through the caudal midbrain, just above the pontomesencephalic junction, clearly shows the destruction of both inferior colliculi.

After surgery the patient was unable to understand verbal communication, but non-verbal hearing, reading, writing, and speaking abilities were unimpaired. He could still identify and localise all sources of non-verbal sounds. Furthermore, he also identified correctly pieces of music that he had known before. Pure tone audiogram and BAEPs were normal and identical to those before operation. Speech audiogram performance, however, had dramatically deteriorated to discrimination scores of 10 and 20.

Hearing impairment related to brainstem disease is rare and the clinical picture is mostly unimpressive with abnormalities detected only by subtle audiological testing. Sixty two cases of hearing impairment by indirect compression of the brainstem in patients with tumours in the pineal region are described in the medical literature. Due to the proximity of the auditory nuclei and their interconnections within the brainstem it was not obvious which auditory centre caused the deficit in these cases. Only three cases had syndromes of pure word deafness with lesions restricted to the inferior colliculi.

It is agreed that only bilateral destruction of the inferior colliculi produces clinical apparent hearing loss. Bognar *et al.*¹ described a patient with a unilateral inferior collicular lesion "with apparent lack of auditory consequences", who exhibited a significant extinction of the contralateral ear during dichotic testing. This extinction might constitute just the mildest degree of failure in verbal comprehension and word deafness, the extreme variant of the same type of auditory disorder. Multiple interconnections at all levels of the brainstem auditory system readily explain the necessity for bilateral destruction to produce apparent clinical consequences.²⁻⁴

It is, however, not easy to explain why the bilateral destruction seen in our patient did not have consequences for higher binaural hearing functions other than linguistic comprehension. Among the important three levels where bilateral interaction primarily occurs—the superior olivary complex, the nuclei of the lateral lemniscus, and the inferior colliculus—the third is the largest com-

ponent and the target of all ascending connections from the lower nuclei. Binaural mechanisms necessary to obtain spatial representation occur essentially at these subthalamic levels. It is known that neurons involved in analysing interaural time or level differences are found abundantly in the central inferior colliculus, but also in subdivisions of the superior olivary complex. Thus an intact superior olivary complex suffices for analysis of interaural time or level differences to localise at least non-verbal sound.

There is evidence that the neurons of the inferior colliculus—by contrast with those of the superior olive—have more specific functions and respond only to interaural intensity changes within a restricted range. This is one argument in favour of the hypothesis that the inferior colliculus possesses "higher" functions than the lower nuclei. The alternate excitatory and inhibitory response of neurons with their ability of dynamic variation of these responses according to different stimulus conditions is the fundamental working principle of the auditory brainstem nuclei. This is a prerequisite for a suggested "gating process" at the inferior colliculus level that may block "irrelevant" sensory input which would be essential for linguistic comprehension. It has been shown that the inferior colliculus incorporates units that respond only to dynamic aspects of a stimulus such as frequency modulated tones in the spoken word.⁴ These physiological aspects could serve as an explanation for the clinical picture in our patient.

The inferior colliculus is thought to be the generator of wave V of BAEPs as documented in numerous human and animal studies.^{1,3} It is well known that a unilateral inferior collicular lesion can be accompanied by normal wave V of BAEPs.¹ Yet completely normal BAEPs in cases of bilateral destruction of the inferior colliculus in humans have not been reported to our knowledge.

We can think of two possible explanations. The first is a technical aspect. As documented in the recent publications of Lapras' group BAEPs are less sensitive in quadrigeminal plate disease than middle latency auditory evoked potentials. Due to the limited experience in this field it can only be speculated whether they might have shown the expected abnormality.¹

On the other hand, some reports postulate an anatomically diffuse second auditory brainstem pathway outside the primary lemniscal projections, called the medial extralemniscal auditory pathway.⁵ Bognar *et al.* favour the existence of this parallel projection system in the "questionable" case of preserved wave V of BAEPs with bilateral inferior collicular destruction.¹

We conclude that our patient allows arguments for the possible existence of a parallel extralemniscal auditory brainstem pathway and a substantial role of the inferior colliculi in the processing of verbal communication.

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Congenital cerebellar ataxia, mental retardation, and atrophic retinal lesions in two brothers

The congenital cerebellar ataxias constitute a rare group of syndromes which are difficult to classify.¹⁻³ Most patients show a non-specific clinical picture characterised by cerebellar ataxia, motor delay, nystagmus, and, often, mental retardation and limb spasticity. The disorder is not progressive and usually improves with age. Autosomal recessive, autosomal dominant, and X linked recessive inheritance have been suggested in different families.³

We recently examined two brothers affected by congenital cerebellar ataxia; they are the only two sons of a healthy white mother who had married her father's brother; the younger sister is apparently normal.

The oldest patient, the first born of three siblings, is 52 years old and in early life showed a mildly delayed psychomotor development. Walking started at about four years of age and ataxia was soon noticed. At the age of 16 he developed severe visual problems because of bilateral keratoconus. At the age of 31, he was admitted to hospital. Hodgkin's lymphoma was detected which responded to chemotherapy. In the past years, cluster headache, memory impairment, vertigo, and mild self injurious behaviour were noted.

He came to our attention because of worsening in memory function. The facies was particular with long ears, downverted palpebral fissures, bulbous tip of the nose, posterior rotation of the ears with adherent earlobes, overcrowded teeth, scrotal tongue, and high arched palate. He had severe visual loss with a deficit of fixation, bilateral rotatory nystagmus, bilateral cataract, gross retinal atrophic lesions, and bilateral dystrophic changes of the iris.

An IQ of 62 was found on the Wechsler adult intelligence scale.⁴ Examination showed cerebellar dysarthria, cerebellar ataxia with limb and truncal ataxia and ataxic gait, mild hypertonia of the lower limbs, hyperexcitable tendon reflexes, and Babinski's sign on the left. Brain MRI showed a pronounced reduction in size of the vermis and of the cerebellar hemispheres and a mild cerebral cortical atrophy (figure).

An ECG was normal and no signs of muscular or peripheral nervous system involvement were found on EMG.

The second patient is a 48 year old man who also showed a moderately delayed psychomotor development. Walking started at about 5 years of age and, also in this case, ataxia was soon noticed. Similarly to his brother, he came under our attention because of worsening in memory function. He had long ears, oval facies and flat occiput, thick lower lip, and dorsal hyperkyphosis. Alternating exotropia, horizontal

T1 weighted midsagittal MR image of the oldest patient (1) in which a severe cerebellar vermis hypoplasia is evident.

nystagmus, dislocation of the left lens in the vitreous humour, and multiple retinal atrophic lesions were also found.

An IQ of 49 was found on the Wechsler adult intelligence scale.⁴ The neurological clinical examination showed cerebellar ataxia with limb and truncal ataxia and ataxic gait; also in this patient, mild hypertonia of the lower limbs, brisk tendon reflexes, and Babinski's sign on the left side were present. Speech was syllabic, poor, and echolalic.

Brain MRI showed a pronounced reduction in size of the vermis and of the cerebellar hemispheres and a mild cerebral cortical atrophy. An EEG was characterised by a slow background activity (6 Hz). No signs of muscular or peripheral nervous system involvement were found on EMG.

Routine blood and urine analysis, lysosomal enzymes, and blood and urinary neurometabolic screening were normal. Karyotype was 46, XY and the molecular analysis for the FMR-1 gene was normal.

These two patients represent a difficult diagnostic problem. They show some common features such as congenital ataxia, nystagmus, mental retardation, and ocular abnormalities (retinal atrophic changes) which make it difficult to include them in one of the groups already proposed.² In particular, the Gillespie syndrome is defined by the association of partial aniridia, congenital cerebellar ataxia, and mental retardation.¹ Our patients were both affected by different ocular abnormalities such as retinal atrophic lesions and nystagmus. Additionally, one of them also had keratoconus, for which surgical treatment was needed and the dystrophic changes of the iris reported above only occurred after the surgical manipulation of the eyes.

For these reasons, we think that our patients are a new example of the considerable clinical and genetic heterogeneity which characterise congenital cerebellar ataxia. Also, the family tree of our patients does not allow us to reach a conclusion on the mode of inheritance of the disorder, even if the strict consanguinity of the parents strongly suggests the possibility of an autosomal recessive inheritance.

It seems important to underline that the patients with congenital ataxia described in the previous literature are usually of a much younger age than our two brothers and that the memory loss reported in the brothers could represent the first symptom of a true dementia process, for which monitoring will be necessary in the future. In fact, even if no assessment of the cognitive status was performed before, our patients had presented a

stable psychomotor picture before the development of memory loss. Thus memory loss and dementia may represent a long term complication of cerebellar ataxia syndromes.

In conclusion, we suggest that our patients may represent a new combination of congenital cerebellar ataxia, mental retardation, and atrophic retinal lesions. This might constitute a new clinical entity if similar patients are found in the future.

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Stimulation single fibre EMG study in a patient with Schwartz-Jampel syndrome

Schwartz-Jampel syndrome is a rare congenital disorder characterised by short stature, oculo-facial abnormalities, bone and joint deformities, clinical myotonia, and persistent spontaneous activity. Lehmann-Horn and colleagues¹ showed two muscle membrane abnormalities in Schwartz-Jampel syndrome by voltage clamp and patch clamp techniques. The abnormalities included reduced Cl^- conductance and synchronised late opening of Na^+ channels. These findings indicate that the origin of the spontaneous activity in Schwartz-Jampel syndrome is located in the muscle membrane itself. However, some reports suggested that the origin of the spontaneous activity may be found in the nerve or end plate because curare seems to decrease or abolish these activities. Single fibre EMG (SFEMG) has been widely employed to study the involvement of nerve or neuromuscular transmission. However, difficulty is anticipated during jitter measurement in Schwartz-Jampel syndrome because the spontaneous involuntary activities may be elicited during voluntary muscle contraction. We employed stimulation SFEMG in a patient with Schwartz-Jampel syndrome in an attempt to obtain a more precise assessment of the neuromuscular transmission. We are unaware of any previous application of this technique in Schwartz-Jampel syndrome.

The patient was a 27 year old woman diagnosed as having Schwartz-Jampel syndrome. She presented clinically with short stature, oculo-facial abnormalities, joint and skeletal deformities, percussion and action myotonia, and hypoaesthetic deep tendon reflexes. Concentric needle EMG (CNEMG) performed when the patient was

13 years old disclosed spontaneous activity evoked by needle movements and muscular contraction that persisted at rest. Neither curare nor lidocaine suppressed the spontaneous activity, but it was suppressed by local ischaemic exercise. The following electrophysiological studies were done: (1) routine nerve conduction studies, including 3 Hz repetitive ulnar nerve stimulation; (2) routine CNEMG; (3) voluntary SFEMG; (4) stimulation SFEMG. Routine nerve conduction studies, repetitive nerve stimulation, and EMG (Counterpoint, Dantec Corp, Denmark) were performed with concentric needle electrodes (Medelec CN-35, UK) and single fibre electrodes (Medelec SF-25, UK). Stimulation SFEMG was performed at the extensor digitorum communis muscle, which showed spontaneous activities without any weakness according to standard methods. Stimulation frequency was set at 5 Hz.

Routine nerve conduction studies that included measurement of distal latencies, conduction velocities, and amplitudes of compound muscle action potentials (CMAPs) and sensory action potentials in upper and lower limbs were normal. Repetitive ulnar nerve stimulation at 3 Hz showed no significant decrement of amplitude and area of CMAPs. CNEMG showed that needle insertion, mechanical stimulation, and mild muscular contraction induced two types of spontaneous activity which persisted even at rest. One type of spontaneous activity was typical myotonic discharges that showed increment and decrement of firing rates and amplitudes. The maximal firing rate was 100 Hz and the duration of the myotonic run was about 10 seconds. Another type of spontaneous activity was high frequency biphasic simple discharges that did not show variations of frequency and amplitudes, and that had a firing rate of about 50 Hz and a duration of 10-30 seconds. Fibrillations were rarely seen. Precise analysis of motor unit potentials (MUPs) were difficult because of interference from spontaneous activities. Among those MUPs measured, the durations were either normal or slightly increased. Findings by SFEMG during voluntary contraction showed that most of the spontaneous activity consisted of single muscle fibre action potentials, different from complex repetitive discharges (CRDs), and frequent occurrence of extradischarges. Although jitter and fibre density measurements by voluntary SFEMG were difficult to perform due to the influence of spontaneous activities, a few jitter values found measurable without interference by spontaneous activity were normal. On stimulation SFEMG, we were able to perform jitter measurement of 21 units, although spontaneous activity induced by electrical stimulation sometimes interfered. On occasions of interference by spontaneous activity, the jitter seemed to be increased. We therefore measured only the periods without spontaneous activity (figure). The mean jitter values ($19.0 \mu s$: normal value at 5 Hz $14.2 (6.6) \mu s$) and individual jitter values were normal except in one end plate.

The electrophysiological studies in the present case of Schwartz-Jampel syndrome are summarised as follows: (a) normal nerve conduction studies; (b) no abnormal decrement by repetitive nerve stimulation; (c) spontaneous activity of typical myotonic discharge and atypical myotonic-like discharge but not CRD, and (d) normal neuromuscular transmission. Based on these electro-