CASE 1996 - 3

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Clinical History:

This boy was delivered by caesarian section for breech presentation at term. Facial asymmetry, torticollis and bilateral congenital hip dislocation were seen soon after birth. Early development was normal but he presented at 21 months with developmental delay, hearing loss and spastic diplegia: developmental assessment indicated 8-9 months delay, brainstem evoked potentials confirmed sensorineural hearing loss. By 2 years there was severe delay, by 3 he was definitely regressing and by 4 he was unable to crawl, with marked spasticity and bulbar palsy. Head circumference remained on the fiftieth centile. Generalised seizures commenced at 5 years and he died of a chest infection at the age of 6. His younger brother presented with hearing loss at 7 months of age. At 4 years old he is now developmentally at a 3 year level and there is significant speech delay.

Necropsy findings:

There was mucopurulent bronchitis and the lung parenchyma was studded with irregular pale firm nodules. Other viscera were unremarkable. The fixed brain was small and weighed 1050g. Coronal slices revealed dilated ventricles, very hard ivory white centra semi-ovalia and cystic disintegrating subcortical U-fibres, thin callosum, soft and greyish-brown basal nuclei and variably thinned cortex. The aqueduct appeared compressed by firm white periaqueductal tissue, the internal structure of pons and medulla was blurred, and the cerebellum showed global folial atrophy, indistinct dentate nuclei and firm greyish white matter.

Material submitted: Frontal lobe section stained with haematoxylin-eosin.