

CASE No 7

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This 20 year old male, who died of aspiration pneumonia, had first come to medical attention at 6 months of age. Following a normal pregnancy and initial postnatal development, he gradually began to demonstrate abnormal motor behaviour, including head nodding, pendular nystagmus and increasing tone in his lower limbs. He never walked and although he could initially crawl using his upper limbs, he lost this ability by 2 years of age. He was never able to speak, but was able to give appropriate yes/no answers to simple questions using his gaze. Throughout his life he was cared for at home by devoted parents. As an adolescent he required intrathecal baclofen for management of spasticity and subsequently G-tube feeding. There was no family history of neurological illness.

Material submitted: Solochrome Cyanine R stained section of the frontal lobe

Question: Diagnosis?