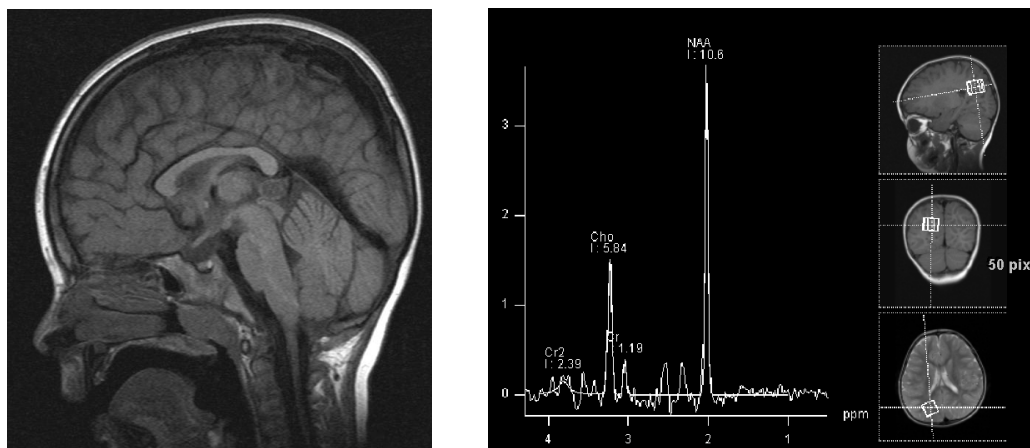


Case study 2 Paediatric: Using spectroscopy to diagnose

Clinical history:

A 3 year old boy presented with inability to walk after a cluster of seizures at age 18 months. Some features of autism were also present with severe learning and communication difficulties. Having been to many other centres for consults and scanning, when he was referred to our hospital we conducted MR spectroscopic examination along with convention MR imaging. The spectroscopy revealed a low peak for a neurotransmitter creatine. Targeted gene testing for its deficiency confirmed creatine transporter defect. The conventional imaging only showed a generalised reduction in the bulk of the cerebral white matter, a non-specific feature.

Results:



Discussion:

This case illustrates well the significant added value of advanced imaging techniques being added on to conventional imaging sequences in difficult cases, where families have journeyed through many hospitals and doctors seeking an answer for their child's developmental issues.

MR spectroscopy is just another imaging sequence which does not need any specific preparation of the patient. When analysed by an expert team, it can provide vital information.