polyminimyoclonus and neurological deficits. Paraneoplastic, autoimmune and neuroinflammatory screen were all unrevealing. The initial electroencephalogram (EEG) was normal; however, the repeat EEG showed focal slowing. Magnetic resonance imaging showed progressive T2 hyperintensity and volume loss predominantly over the left frontoparietal lobes, but also involved the right frontal lobe. Brain biopsy showed features of a T-cell predominant encephalitis. Rasmussen's encephalitis was suspected and immunotherapy with steroids and intravenous immunoglobulin were initiated, but he continued to deteriorate. He is now started on cyclophosphamide.

Conclusions Our case adds to the accumulating number of cases of A-RE reported in the literature. The rarity of the condition and overlap in clinical phenotype with other central nervous system diseases lead to delayed diagnosis in almost all reported cases. In our patient, the bilateral MRI changes and delayed recognition of seizures compounded to the diagnostic uncertainty. The main therapeutic options in adults are antiepileptic and immunomodulatory agents, and whilst there is no cure, early and aggressive therapy may result in improved patient outcomes.

2284 A CASE OF NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA) PREVIOUSLY DIAGNOSED AS SPASTIC DIPLEGIC CEREBRAL PALSY

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Objectives To present an 'ultrarare' case of neurodegeneration with brain iron accumulation (NBIA) that was previously diagnosed as spastic diplegic cerebral palsy.

Background NBIA is a very rare condition and affects both children and adults. It is clinically and genetically heterogenous and is characterized by pathological brain iron accumulation in the basal ganglia. Neurological manifestation include progressive extrapyramidal syndrome, pyramidal signs, cognitive dysfunction or ocular abnormalities. Neuroimaging shows T2 hypointense lesions bilaterally in the globus pallidi and substantia nigra on T2 weighted images

Case This is a case of a 24year old male who was diagnosed by paediatricians as 'cerebral palsy spastic diplegia' with intellectual disability. He was born at term and met all developmental milestones till age 3. He then had difficulties with gait, incoordination and delayed speech. He was diagnosed as a child as spastic diplegic cerebral palsy. His mobility declined further and he was assessed in adult neurology clinic for seizures. MRI brain, previously normal as a child, showed changes consistent with neurodegeneration with brain iron accumulation in the substantia nigra and atrophy of corpus collosum and cerebellum. Genetic testing confirmed heterozygous pathogenic mutations for PLA2G6 gene, securing diagnosis of phospholipase A2-associated neurodegeneration (PLAN).

Conclusion This case offers a reminder to re-evaluate diagnoses when clinical course of disorder does not fit the known pattern. Distinction between static and progressive clinical course is crucial. Regression in development should alert clinicians to metabolic and neurodegenerative disorders. NBIA, although 'ultrarare', should be considered as a differential if phenotypically appropriate.

2285 EFFICACY AND SAFETY OF AVALGLUCOSIDASE ALFA IN PARTICIPANTS WITH LATE-ONSET POMPE DISEASE AFTER 97 WEEKS OF TREATMENT DURING THE COMET TRIAL

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Objectives Report efficacy/safety of avalglucosidase alfa (AVAL) in participants with late-onset Pompe disease in the extended treatment period (ETP) of Phase 3 COMET(NCT02782741) after a 49-week primary analysis period (PAP).

Methods At PAP enrollment, participants were treatment-naïve (n=100; age 16–78 years). All 51 participants receiving AVAL 20mg/kg every 2 weeks (qow) in the PAP continued this in the ETP (AVAL-arm). Of 49 participants receiving alglucosidase alfa (ALGLU) 20mg/kg qow in the PAP, 44 entered the ETP switching to AVAL 20mg/kg qow (Switch-arm).

Results Trends for improvement or stabilization from Baseline to Week 97 were observed for the primary and secondary outcomes of respiratory and motor function. Changes (LSmean[SE]) in forced vital capacity%predicted: AVAL-arm, +2.65[1.05]; Switch-arm, +0.36[1.12] and 6-minute walk test distance: AVAL-arm, +18.60[12.01]m; Switch-arm, +4.56[12.44]m. Similar trends occurred for muscle strength and quality of life measures. Treatment-emergent adverse events (AEs) in the ETP occurred in 49(96%) AVAL-arm and 42(95%) Switch-arm participants. Five discontinued during the ETP due to 6 treatment-emergent AEs; 4 were treatment-related (ocular hyperemia, erythema, urticaria, respiratory distress) and 2 were non-treatment-related (acute myocardial infarction, pancreatic adenocarcinoma). In the ETP, 12 AVAL-arm and 10 Switch-arm participants had treatment-emergent serious AEs (SAEs); 3 and 2 of them, respectively, had treatment-related SAEs. Switch-arm participants showed no safety- or immunogenicity-related concerns.