Radiological and Laboratory Assessment of Children with Progressive Encephalopathy in Fayoum University Hospitals

Abstract:

Background: Progressive neurological disease in children poses an important challenge to health systems in terms of diagnosis and management. Progressive Encephalopathy (PE) is often used interchangeably with neurodegenerative encephalopathy. Both terms lack a firm definition, but PE is preferred because it encompasses clinically progressive conditions without demonstrable neuronal loss as well as those with a demonstrable loss of neural tissue. They are often detected by magnetic resonance imaging (MRI) examination. **Objectives:** The study aimed to determine the value of different radiological and laboratory studies as diagnostic tools for different etiologies of progressive encephalopathy (PE) and to investigate the relation between them. The study also aimed to illustrate the prevalence of different etiologies of PE among outpatient clinic attendants. **Methods**: Our study is a cross-sectional descriptive study. It included 79 patients aged between 3 months old up to 12 years old who sought medical advice at Neuropediatrics Clinic, Fayoum University Hospitals. They presented with progressive alteration of mental status with/without motor affection during a period of 18 months from December 2016 till June 2018 and underwent clinical, laboratory, radiological and neurophysiological assessment. **Results:** Out of 79 cases showing manifestation of Progressive Encephalopathy: A-15 cases (19%) were diagnosed as Neurocutaneous diseases. B-Inherited metabolic diseases (IMD) represented 55 cases (70%) .C- Epileptic syndromes were 4 cases (5%) (2 cases of Dravet syndrome and 2 cases of West syndrome). Five cases were undiagnosed and need molecular testing. The most common cause of PE was inherited metabolic diseases (70%). Neurocutaneous disorders accounted for 19%. The early age of presentation strokes alarms for inherited metabolic diseases as neurometabolic and urea cycle diseases (17/18 cases of this group were diagnosed before 2 years of age and 7/18 cases were diagnosed during neonatal period), mitochondrial diseases (100% of cases during infancy), white matter diseases (5/7 cases diagnosed during infancy except for Van der Knaap and adrenoleukodystrophy). The late age of manifestations rises susceptibility of neurocutaneous disorders and Wilson disease as well. Gaucher disease can be diagnosed at any age. **Conclusion**: Careful Examination is very important in all cases of PE. Imaging studies can be the next step to the diagnosis in cases of neurocutaneous syndromes. MRS is helpful for diagnosis of mitochondrial diseases as well as Canavan disease. Neurometabolic diseases can be diagnosed by laboratory and enzyme assay. Neonatal screening programs can spare time and effort. Molecular diagnosis can be a helpful diagnostic tool in case of doubtful or undiagnosed cases. Epileptic syndromes can be diagnosed clinically together with EEG (west syndrome) and by molecular diagnosis. Undiagnosed cases need molecular testing for diagnosis.