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

Thesis
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Summary

Progressive neurological disease in children poses an important challenge to health systems in terms of diagnosis and management.

The term encephalopathy describes a diffuse brain disorder of the brain in which at least two of the following symptoms are present: (1) altered conscious state ;(2) altered cognition or personality; (3) Seizures.

Progressive Encephalopathy 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, most often detected by magnetic resonance imaging (MRI) examination.

This 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 neurology 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.

The study aimed to evaluate different patterns of clinical presentations and to determine the value of different radiological and laboratory studies as diagnostic tools for different etiologies of progressive encephalopathy (PE). It also aimed to correlate between radiological and laboratory findings.

In our study, cheap diagnostic tools were the main cornerstone for diagnosis. Being a developing country, it is not easy to ask for molecular diagnosis once you need and you should ask for cheaper tools.

Clinical diagnosis was very important in all cases and was enough for diagnosis of Neurocutaneous Syndromes (15/79 cases were clinically diagnosed with no need for other confirmatory tests) .Imaging can be the next step to confirm diagnosis if needed.

Inhreited Metabolic diseases (55/79 cases) were diagnosed by laboratory and enzyme assays. Clinical diagnosis is not enough in this group.

3 cases were diagnosed by neonatal screening programs (2 PKU cases and one case of GA) sparing time and effort. One case of biotinidase deficiency was diagnosed by enzyme assay together with 13 cases of storage diseases.

Neurometabolic and Urea cycle disorders (18/79 cases) were diagnosed by TMS/MS as well as OAU. Brain imaging as in GA can be helpful but not enough for diagnosis in this group.

Neuroimaging was helpful in cases of white matter involvement as Canavan disease (MRS was confirmatory), Leukodystrophies as Metachromatic, Krabbe and Adrenoleukodystrophy were confirmed by enzyme assay later on. Van der knapp disease was confirmed by MRI with no need for further assessment.

Mitochondrial diseases (14/79 cases) can be diagnosed by combination of clinical, laboratory (S.Lactate and S.CK), Brain imaging (MRI and MRS) and electrophysiological findings (EMG) .For diagnosis of Leber disease , VEP and ERG can also be needed.

Molecular diagnosis was done in all 5 Gaucher cases and two Neimann pick cases. One Neiman pick type C was diagnosed after misinterpretation as a Gaucher case by Molecular techniques.

Epileptic syndromes were diagnosed clinically together with EEG (west syndrome) and one case was diagnosed as SCN1 related disorder by molecular diagnosis

Miscellaneous cases need molecular testing for diagnosis.