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Chromatographic detection of inborn error of metabolism in Egyptian pediatrics, two years of experiences

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Background: Inborn errors of metabolism (IEM) represent a special challenge in pediatric practice. Newborn screening approaches by tandem mass spectrometry (MS/MS) coupled to gas chromatography (GC) early in infancy help in rapid and well-timed therapeutic interference to prevent overwhelming neurological outcomes.

Aim: Aim of this study is to estimate the effectiveness of the metabolic alterations as a rapid non-invasive metabolomics screening technique for diagnosis of different type of IEMs to establish diagnostic clue to IEM in high risk Egyptian pediatric for proper treatment and better outcome.

Methods: During 2015-2016, samples of 480 patients were analyzed. Quantitative measurements of amino acids and acylcarnitine profiles using MS/MS and of organic acids using GC/MS were done.

Results: 39 (39/480, 8.1%) of the patients were diagnosed to have IEM. The following disorders were identified; organic acidopathies: 25 (64.1%), amino acidopathies: 9 (23.1%), fatty acid disorders (FAO): 3 (7.7%) and lactic academia (LA): 2 (5.1%).

Conclusion: Newborn screening program should be established in Egypt as the overall incidence of IEM was found to be high due to raised ratio of consanguinity; this approach could prevent, or at least, reduce serious neurological and developmental sequelae, improve survival and reduce mortality and morbidity of patients.

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Chemical composition of medicinal plant of Atractylis genus

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The research of active principles extracted from the aromatic and medicinal plants is of a key importance, because it allows the development of drugs to maintain the health of the human being. It is for this reason that we are interested in this type of investigation in our research laboratory. Besides, our laboratory has recently started a research program intended for the systematic study of medicinal plants belonging to the genus Atractylis (Asteraceae family) in view of their valorization. The present study focuses mainly on the species *Atractylis cancellata*. The Asteraceae family is characterized by its wealth in secondary metabolites of biological interest such as the triterpenes, the phenolic compounds of type flavonoids, and sesquiterpenes. The plants of the genus Atractylis are deemed in traditional medicine for treating many diseases. The main purpose of our work is to isolate and identify secondary metabolites that may have a biological activity from the different organic extracts of the species *A. cancellata*. The hydroalcoholic extraction: alcohol/water, separation and purification chromatographic: VLC, CC and TLC of extracts petroleum ether and ethyl acetate acquired from the whole plant *A. cancellata*, allowed us to isolate and identify secondary metabolites of different types flavonoids, sterols and triterpenes. The determination of their molecular structures is performed by the spectroscopic analysis methods such as the NMR 1D (1H, 13C J-modulated and DEPT) and the NMR 2D (COSY, HSQC, HMBC, and NOESY) and the mass spectrometry ESI-MS. It is mainly of lupeol, oleanolic acid, β -sitosterol and apigenin.

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