High-risk babies or children from regional hospitals are routinely referred to the Metabolic Division of Child Health Department at SQUH.ESI-MS/MS was carried out on a Quattro Micro (Manchester, UK), triple quadrupole spectrometer, equipped with a Waters Alliance 2790 HPLC pump, with minor modification of methods developed by Chance et al.1–2 and Rashed et al.3 A 3 mm disk from a selected spot from the Guthrie card was punched out using a standard hole punch in each of a 96-well polypropylene microtitre plate.If the second sample agreed with the first sample result, the diagnosis was then confirmed by urine organic acids analysis, plasma amino acids analysis, or direct enzyme assay; all analyzed by Biomnis laboratory, France (BIOMNIS Specialised Medical Pathology, International Division, Lyon Cedex 7 – FRANCE).Internal standards are represented with a small superscript star In Electrospray-ionization tandem mass spectrometry (ESI- MS/MS), a sample is first dissolved in a mixture of water and or- ganic solvent (e.g., acetonatrile).The sample in the blood spot punch in each well was then extracted by adding 100 ul of isotope- labeled internal standard solution of amino acids and acylcarnitines (Cambridge Isotope Labs, Andover, MA, USA) and incubated at room temperature for 30 minutes.Whole blood samples were drawn by heel prick or venipuncture from high-risk babies and spotted on Guthrie filter cards No. 903 Whatman paper (Schleicher and Schuell, Dassel, Germany).The Omani population was divided into three ethnic groups: Arabs of the north, Arabs of the south (Dhofari), and Omanis of Asian descent.Ionization of the derivatized analyte is achieved by electro- spray ionization in which the sample effluent is passed through a small capillary to which a high voltage has been applied.Ions are separated by electromagnetic fields, detected by a quantitative method and processed into mass spectra (Fig.The vertical axis represents ion intensity expressed as a % of the largest peak in the spectrum.Then, the extract is derivatized with butanol to form butyl esters of amino acids, organic acids, and fatty acids.The ioniza- tion process involves the transfer of a charge to the solvent droplets, evaporation of the solvent, and finally, production of positively and negatively charged ions.Neolynx operates by calculating the ratio of the intensity of the mass spectral peak for the analyte to the intensity of the mass spectral peak for a pre-defined internal standard.Omanis of Asian Origin included migrants from Iran, India, Pakistan and Afghanistan.This vaporized and ionized mixture enters the MS 1, which functions as a separating device and allows only the ion(s) of interest to pass through.Since certain compound classes share common fragment ions of neutral fragment molecules, a special analysis can be set up to only detect precursor ions with a particular fragment.???????????

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