

Resource Summary Report

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International Database of Tetrahydrobiopterin Deficiencies

RRID:SCR_008171

Type: Tool

Proper Citation

International Database of Tetrahydrobiopterin Deficiencies (RRID:SCR_008171)

Resource Information

URL: <http://www.bh4.org/BH4DatabasesBiodef.asp>

Proper Citation: International Database of Tetrahydrobiopterin Deficiencies (RRID:SCR_008171)

Description: THIS RESOURCE IS NO LONGER IN SERVICE, documented on August 26, 2016. The BIODEF database have tabulated the most common clinical and laboratory data related to hyperphenylalaninaemia and tetrahydrobiopterin deficiencies. Additionally, there are data regarding treatment, outcome, and DNA analysis. Approximately 2% of newborns with hyperphenylalaninaemia are deficient in tetrahydrobiopterin. Selective screening must be performed in all instances where hyperphenylalaninaemia is detected by neonatal screening. In the last 20 years, 308 patients with tetrahydrobiopterin deficiencies have been recognized as a result of screening carried out, worldwide, in Departments of Paediatrics. Of these 308 patients, 181 suffered from 6-pyruvoyltetrahydropterin synthase deficiency, 92 from dihydropteridine reductase deficiency, 13 from pterin-4a-carbinolamine dehydratase deficiency, 12 from GTP cyclohydrolase I deficiency, and 10 are still unclassified. The BIODEF database have tabulated the most common clinical and laboratory data related to hyperphenylalaninaemia and tetrahydrobiopterin deficiencies. Additionally, there are data regarding treatment, outcome, and DNA analysis. Preliminary evaluation reveals that the degree of hyperphenylalaninaemia can vary from normal to 2500 $\mu\text{mol/L}$. Analyses of pterins in urine and measurement of dihydropteridine reductase activity from Guthrie cards are absolutely essential tests for accurate diagnosis. There is a regional (demographic) variation in the frequency of tetrahydrobiopterin deficiencies indicating the highest incidence in Saudi Arabia, probably a consequence of the high consanguinity rate.

Synonyms: BIODEF

Resource Type: database, data or information resource

Keywords: ethnic, frequency, 6-pyruvoyltetrahydropterin synthase deficiency, analysis, bh4, clinical, deficiency, demographic, diagnosis, dihydropteridine reductase deficiency, dna, gtp cyclohydrolase i deficiency, hyperphenylalaninaemia, measurement, neonatal, origin, outcome, pterin, pterin-4a-carbinolamine dehydratase deficiency, sex, tetrahydrobiopterin, treatment, urine

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: International Database of Tetrahydrobiopterin Deficiencies

Resource ID: SCR_008171

Alternate IDs: nif-0000-21052

Record Creation Time: 20220129T080245+0000

Record Last Update: 20260808T190420+0000

Ratings and Alerts

No rating or validation information has been found for International Database of Tetrahydrobiopterin Deficiencies.

No alerts have been found for International Database of Tetrahydrobiopterin Deficiencies.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We have not found any literature mentions for this resource.