Creatine and guanidinoacetate reference values in a French population

Submitted by a.bergoend on Thu, 04/30/2015 - 10:42

Titre Creatine and guanidinoacetate reference values in a French population

Type de publication Article de revue

Auteur Curt, Marie Joncquel-C [1], Cheillan, David [2], Briand, Gilbert [3], Salomons, Gajja S [4], Mention-Mulliez, Karine [5], Dobbelaere, Dries [6], Cuisset, Jean-Marie [7], Lion-François, Laurence [8], Portes, Vincent Des [9], Chabli, Allel [10], Valayannopoulos, Vassili [11], Benoist, Jean-François [12], Pinard, Jean-Marc [13], Simard, Gilles [14], Douay, Olivier [15], Deiva, Kumaran [16], Tardieu, Marc [17], Afenjar, Alexandra [18], Héron, Delphine [19], Rivier, François [20], Chabrol, Brigitte [21], Prieur, Fabienne [22], Cartault, François [23], Pitelet, Gaëlle [24], Goldenberg, Alice [25], Bekri, Soumeyla [26], Gerard, Marion [27], Delorme, Richard [28], Porchet, Nicole [29], Vianey-Saban, Christine [30], Vamecq, Joseph [31]

Editeur Elsevier

Type Article scientifique dans une revue à comité de lecture

Année 2013

Date Jan-11-2013

Numéro 3

Pagination 263-267

Volume 110

Titre de la revue Molecular Genetics and Metabolism

ISSN 10967192

Mots-clés Creatine [32], Creatinine [33], Guanidinoacetate [34], Laboratory values [35], Plasma [36], Urine [37]
Creatine and guanidinoacetate are biomarkers of creatine metabolism. Their assays in body fluids may be used for detecting patients with primary creatine deficiency disorders (PCDD), a class of inherited diseases. Their laboratory values in blood and urine may vary with age, requiring that reference normal values are given within the age range. Despite the long known role of creatine for muscle physiology, muscle signs are not necessarily the major complaint expressed by PCDD patients. These disorders drastically affect brain function inducing, in patients, intellectual disability, autistic behavior and other neurological signs (delays in speech and language, epilepsy, ataxia, dystonia and choreoathetosis), being a common feature the drop in brain creatine content. For this reason, screening of PCDD patients has been repeatedly carried out in populations with neurological signs. This report is aimed at providing reference laboratory values and related age ranges found for a large scale population of patients with neurological signs (more than 6 thousand patients) previously serving as a background population for screening French patients with PCDD. These reference laboratory values and age ranges compare rather favorably with literature values for healthy populations. Some differences are also observed, and female participants are discriminated from male participants as regards to urine but not blood values including creatine on creatinine ratio and guanidinoacetate on creatinine ratio values. Such gender differences were previously observed in healthy populations; they might be explained by literature differential effects of testosterone and estrogen in adolescents and adults, and by estrogen effects in prepubertal age on SLC6A8 function. Finally, though they were acquired on a population with neurological signs, the present data might reasonably serve as reference laboratory values in any future medical study exploring abnormalities of creatine metabolism and transport.

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DOI 10.1016/j.ymgme.2013.09.005 [39]
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