Gaucher Disease: Multidisciplinary Approaches and Therapeutic Advances: Experience of a Neuropediatrics Department in Morocco

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Abstract :

Gaucher disease is a rare lysosomal disease that is curable and characterized by clinical polymorphism. We report the experience of the Neuropediatrics Department in Rabat concerning 17 pediatric cases of Gaucher disease collected over a 5-year period, with a specific focus on clinical, paraclinical, therapeutic, and evolutionary features.

Our series includes 17 patients, comprising 9 boys and 8 girls, with an average age of 10 years and 6 months at the start of the study. Five of our patients come from consanguineous marriages. The identified phenotypes are as follows: type 1 in 13 patients, type 3 in 3 patients, and type 2 in a single patient. The characteristic clinical presentation is mainly represented by the triad of hepatosplenomegaly, hemorrhagic syndrome, and bone symptoms. Neurological signs were present in 23% of our patients. Fourteen patients received specific treatment for Gaucher disease, while three passed away during the follow-up period. The treatments received by our patients were as follows: imiglucerase in 8 cases, velaglucerase in 5 cases, and taliglucerase in one case. No allergic reactions were recorded. Regular monitoring was ensured through clinical, hematological, radiological assessments, and biomarker research. Some of our patients participated in providing insights into the value of lyso-Gb1 as a predictive biomarker in treatment-naïve patients with Gaucher disease type 1 in the Lyso-Proof study[4].

Our longitudinal analysis also highlights the effectiveness of available treatments but also underscores the importance of early diagnosis and continuous monitoring to improve the quality of life and survival of patients. This experience contributes to a better understanding of the disease's progression and the optimization of care for children with Gaucher disease.

RECENT PUBLICATIONS :

1. The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here? Neal J. Weinreb a, Ozlem Goker-Alpan Volume 136, Issue 1, May 2022, Pages 4-21
2. Diagnosis features of pediatric Gaucher disease patients in the era of enzymatic therapy, a national-base study from the Spanish Registry of Gaucher Disease . Published online 2017 May 3.
3. Pediatric Gaucher Disease Presenting with Massive Splenomegaly and Hepatic Gaucheroma Giuanluca bossu *Children* 2023, *10*(5), 869;
4. Insights into the Value of Lyso-Gb1 as a Predictive Biomarker in treatment-Naive patients with Gaucher Disease Type 1 in the LYSO-PROOF Study. Filipa Curado. Diagnostics 2023, 13, 2812
5. Pharmacological treatment of pediatric Gaucher disease. Punita Gupta Expert Review of Clinical Pharmacology Volume 11, 2018 - Issue 12



Dr. Yamna Kriouile, a neuropediatric specialist and professor of pediatrics, heads the pediatric endocrinology and neurology department, specializing in inborn metabolic disorders.

With extensive experience in teaching and research evaluation administration across esteemed healthcare institutions and educational establishments, Dr. Kriouile's career boasts remarkable contributions to clinical research and rare metabolic disease patient care. Her work has elucidated intricate disease mechanisms, paving the way for novel therapies.

Through her role as the vice president of the Moroccan Society for study of Inborn Errors of Metabolism (SSIEM) and Moroccan Association of Neuropediatrics, she is experienced in admnistration, and public relations.

As a professor in higher education , she has positively influenced healthcare professionals, inspiring their commitment to improving care for rare metabolic disorder patients. Fueled by a passion for medical innovation and an unwavering dedication to enhancing patients' quality of life, she is a prominent figure in neuropediatrics and rare metabolic diseases.

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