

RARE DISEASES AND HANDICAP: State : médical, social, health policy in developing countries. Case of Cameroon: about a case: Cushing's disease : Review

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Uncommon and orphan diseases are a reality throughout the sector and a real public health trouble in developing international locations. The superb precariousness wherein the populations stay will increase their impact and their gravity by means of the absence of statistics, technical platform, manner of detection, real presence of research and medical research on these territories, lack of information of the detection, diagnosis, with out forgetting that the way of care and prevention are from time to time inexistent or little acknowledged and specifically pricey in nations in which the populations are terrible, with out mutual or social protection as in industrialized nations . rare issues represent a big and heterogeneous group of diagnoses of which many motive chronic disabilities with widespread impact at the lives of affected individuals and their families in addition to on the fitness-care device. every individual ailment is rare, however while considered as a set, rare disorders are not unusual with a complete prevalence of about 6-8%. The scientific presentation of these issues includes a large variety of symptoms and signs and symptoms, regularly concerning the anxious device and ensuing in signs such as high brow disability, neuropsychiatric disorders, epilepsy and motor dysfunction. The techniques for establishing an etiological analysis in patients with uncommon problems have progressed dramatically all through latest years. With the advent of genomic screening methods, it's been shown that the cause is genetic in most of the people of the patients and lots of will obtain an etiological diagnosis in a scientific putting. but, there are numerous challenges in diagnosing these problems and in spite of current years' advances, a large wide variety of patients with rare disorders still cross with out an etiological prognosis. on this chapter we will review the etiology of uncommon issues with consciousness on high brow disability and what has been learned from large parallel sequencing studies in deciphering the genetic basis. furthermore, we are able to speak challenges in the etiological diagnostics of those problems including troubles that regard interpretation of the severe genetic editions detected by way of genomic screening techniques and demanding situations inside the translation of massive parallel sequencing technologies

into scientific exercise. The country wide foundation of Dentistry for the Handicapped (NFDH) is a national charitable business enterprise entirely dedicated to meeting the wishes of residents with bodily, clinical, and intellectual disabilities via presenting dental care to disabled and elderly those who cannot have enough money remedy and are not eligible for public aid. Our united states' trendy of dental care is one of the maximum inside the global, yet those individuals maximum desperately in need regularly pass untreated. they'll be children or adults who are mentally or physically disabled. They suffer from cerebral palsy, more than one sclerosis, mental illness, and many different disabilities. the inspiration was mounted in 1974 and is an affiliate of the yankee Dental affiliation and state and nearby dental associations. Disabled, aged, and mentally compromised sufferers are connected with dentists of their communities to get hold of loose complete dental remedy, together with prosthetics. there's also a application to offer education to help mother and father and professionals being concerned for developmentally disabled individuals to provide appropriate oral hygiene. The countrywide corporation for rare issues (NORD) net website online, its databases, and the contents thereof are copyrighted by NORD. No a part of the NORD net website, databases, or the contents can be copied in any manner, including but no longer constrained to the following: electronically downloading, storing in a retrieval gadget, or redistributing for any business functions with out the explicit written permission of NORD. Permission is hereby granted to print one tough replica of the data on an character's sickness for your non-public use, furnished that such content material is in no way modified, and the credit for the supply (NORD) and NORD's copyright note are blanketed at the revealed reproduction. any other electronic duplicate or different revealed variations is strictly prohibited. This work will consciousness on a patient case supplying a rare ailment in this case Cushing's disorder. A case that occurred in Cameroon for you to set up in a realistic manner this predominant and yet overlooked, neglected issue in a fitness system with terrible specialised systems, technical structures, and without the

support of the pharmaceutical laboratories that would with the health machine of these bad nations set up a system of refueling inside the experience of the social view the feathat calls for the monitoring of these lengthy pathologies that make self sufficient living almost not possible, also lifestyles-threatening. In Cameroon, there had been problems in the care and monitoring of this younger affected person due to the fact adolescence thru the section of the pubertal transition, adolescence, to maturity. it's far consequently vital to organize the follow-up of these sufferers, by means of developing particular applications of medical follow-up, psychological care, and social integration applications health policies need to invest in higher aid.

BIOGRAPHY

Dr. Carolle-Laure KPOUMIEMD , phd in public health and social medicine, has worked as pediatric assistant physician in pediatric's department of faculty of medicine in Cameroon, as clinical researcher, scientific writing, public health, oncology, rare diseases, orphan drugs, handicap, internal medicine and continued her career in France. She participates in many congresses with studies made in hospitals on relevant cases in the framework Studies and clinical research. She became head of department, director, where she continued her research, writing of treatment protocols in different departments for common pathologies encountered in her context, managing Epidemic crisis , rare diseases, participates in studies on neonatal mortality in underdeveloped countries. She work on ADR in hospital following drug administration, pharmacovigilance, vaccinovigilance, biovigilance, materiovigilance, pharmacovigilance in underdeveloped countries, articles, medical information and therapeutics safety, clinical trials, Post-marketing, and other's studies.

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