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In this issue

Research Article

Open Access Research Article PTZAID:GJRD-5-126

A multidisciplinary clinic for Filipino patients with skeletal dysplasia: Opportunities and Challenges

Published On: October 24, 2020 | Pages: 027 - 029

Author(s): Maria Melanie Liberty B Alcausin, Ebner Bon G Maceda*, Gracia Cielo E Balce, Joycie Eulah H Abiera and Maria Glorian B Tomen

Purpose: Skeletal dysplasias comprise a heterogenous group of genetic disorders that have generalized abnormalities in cartilage and bone. Although individually rare, collectively it is common with an estimate of 1 in 2000 to 3000. Individuals with skeletal dysplasias are known to be at risk for a myriad of medical complications associated with their conditions; hence ...

Abstract View Full Article View DOI: 10.17352/2640-7876.000026

Review Article

Open Access Review Article PTZAID:GJRD-5-128

A Review on Psychological and Socio-Economic Impacts of Corona Virus Disease (Covid-19) The Case of Under Developing Countries

Published On: November 20, 2020 | Pages: 036 - 039

Author(s): Desalegn Wondim Alene*

A history signifies that there were different pandemic diseases across the globe at different times that brings a fundamental consequence in psychological, socio-economic and political situations. Evidences showed that most emergency diseases were geographically restricted while others were/are not that attracts the professionals in developing insight and taking cross ...

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Open Access Review Article PTZAID:GJRD-5-119

The case number 52 of Ruprecht Majewski-Bosma syndrome associated with atrial septal defect

Published On: February 28, 2020 | Pages: 001 - 003

Author(s): Aamir Jalal Al Mosawi*

Background: Ruprecht Majewski-Bosma syndrome is an extremely rare dysmorphic syndrome results from severe hypoplasia of the nose and eyes occurring in association with palatal abnormalities. It is characterized by congenital complete nasal agenesis (Bilateral aplasia of the nose), microphthalmia including clinical anophthalmia, hypertelorism and other eye defects, hig ...

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Case Report

Open Access Case Report PTZAID:GJRD-5-127

Mitochondrial disease, hypertrophic cardiomyopathy and cutaneous lupus in an infant with food hypersensitivity

Published On: November 04, 2020 | Pages: 030 - 035

Author(s): E Estrada-Reyes*, D Lopez-Gallegos, E Faugier-Fuentes, M Pardo-Castañeda, E Barragán- Perez, I Nuñez-Barrera, M Sanchez-Ruiz, S Nuñez-Barrera and G Ramon-Garcia

This paper describes a 4 month-old-girl with food allergies, mitochondrial disease, cutaneous lupus, and hypertrophic cardiomyopathy. She suffered from infectious pericarditis due to Coxsakie virus as a complication. She additionally presented bicytopenia (hemoglobin levels 8.9 g/dL, platelets 127 x 103 / µL), high lactate levels (5mmol/L), seizures, hypertrophic card ...

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Open Access Case Report PTZAID:GJRD-5-125

Temporary improvement of motor symptoms of a patient with Parkinson’s disease after accidental electric shock - Case report

Published On: September 09, 2020 | Pages: 025 - 026

Author(s): Antônio Marcos Da Silva Catharino, Kattiucy Gabrielle Da Silva Brito, Edarlan Barbosa Dos Santos, Gilberto Canedo Martins and Marco Antonio Orsini Neves

Despite parkinson's disease to be one of the most frequent movement disorders, with motor and non-motor symptoms and pharmacological and surgical treatments, we present a case of temporary improvement of motor symptoms after an accidental electric shock and we highlight the need for furthrer studies to discuss possible mechanisms involved in this case. ...

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Open Access Case Report PTZAID:GJRD-5-124

Duane-Radial Ray syndrome a SALL4-Related Disorder. Report of a case in Chile

Published On: September 07, 2020 | Pages: 022 - 024

Author(s): Jonathan Huserman* and Catherine Diaz

The Duane-Radial Ray syndrome or Okihiro syndrome belongs to the SALL4-Related Disorders, a phenotypic spectrum, that additionally includes, acrorenoocular syndrome and Holt-Oram syndrome, caused by the alteration of the same gene, which has significant relevance in the mesoderm, the limbs, and the heart development. These syndromes are characterized by thumb alterati ...

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Open Access Case Report PTZAID:GJRD-5-122

Intrahepatic Cholangiocarcinoma Skin Metastasis in a Patient with Hidradenitis Suppurativa: A Rare Entity

Published On: April 24, 2020 | Pages: 010 - 014

Author(s): Ali Y Fakhreddine*, Theresa Yang, Guanghong Liao and Bahman Chavoshan

We present a challenging diagnosis of disseminated intrahepatic cholangiocarcinoma presenting with perineal cutaneous masses in a young African American male with known hidradenitis suppurativa. The patient was a 39-year-old male who presented to the emergency department with difficulty walking due to severe gluteal swelling and pain. The patient had an 18-month histo ...

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Open Access Case Report PTZAID:GJRD-5-121

Hematuria in a Patient with Non-malignant Bladder Nodules

Published On: April 13, 2020 | Pages: 007 - 009

Author(s): Terese S Bergheim, Visal Nga, Andrew C Burg and Bahman Chavoshan*

Introduction: Gross hematuria in adults with resultant anemia is highly concerning for genitourinary malignancy. However, in rare instances, malakoplakia can mimic such malignancy. Malakoplakia is a benign granulomatous condition with malfunction of the phagolysosomal activity of macrophages and monocytes. This leads to formation of foamy histocytes with intracytopla ...

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Open Access Case Report PTZAID:GJRD-5-120

Cutis marmorata telangiectasia congenita-a needle in the neonatal dermatology haystack?

Published On: April 03, 2020 | Pages: 004 - 006

Author(s): Hassan Shakeel* and Ather Ahmed

Cutis Marmorata Telangiectasia Congenita (CMTC, also known as van Lohuizen syndrome) is a rare disorder characterised by dilatation of the cutaneous vasculature. This results in a blue-purple ‘marbled’ appearance of the skin due to telangiectasia, phlebectasia and persistent cutis marmorata. It is often mistaken for benign cutis marmorata and is therefore likely under ...

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Case Study

Open Access Case Study PTZAID:GJRD-5-123

Double homozygous Cystic Fibrosis Transmembrane Regulator gene (CFTR) mutation: A case series and review of the literature

Published On: August 18, 2020 | Pages: 015 - 021

Author(s): Hanaa Banjar*, Wesam Alkassas, Firas Ghomraoui, Reem Ghomraoui and Nabil Moghrabi

Introduction: Double homozygous mutation with the presence of double mutations in each allele is a very rare

phenomenon with only 2 reports that have described this phenomenon in the medical literature. Objective: To find the prevalence of double homozygous in our Cf population and to describe their mutations and review of the literature in this phenomenon. Methodol ...

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