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

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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 ...

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

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

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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 ...
Hematuria in a Patient with Non-malignant Bladder Nodules

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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 ...

Abstract View  Full Article View  DOI: 10.17352/2640-7876.000021

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

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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 ...

Abstract View  Full Article View  DOI: 10.17352/2640-7876.000020

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

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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...