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

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

Author(s): Terese S Bergheim, Visal Nga DO, 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 intracytoplasm ... 

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 ...