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A recognizable type of pituitary, heart, kidney and skeletal dysplasia mostly

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1,560 条结果 时间不限

Skeletal Dysplasia, Global Developmental Delay, and ... 翻译此页

Skeletal Dysplasia, Global Developmental Delay, and Multiple Congenital Anomalies in a 5 year-old boy– Report of the Second Family with B3GAT3 mutation and Expansion of the Phenotype
https://www.researchgate.net/publication/261105147_**Skeletal_Dysplasia**_Global...

Anterior Pituitary Failure - ScienceDirect 翻译此页

Cited by: 11 Author: John D. Carmichael
Publish Year: 2011

Anterior pituitary failure is caused by several etiologic factors. Mass lesions and their treatment, genetic mutations, infiltrative and infectious disease, and traumatic brain injury are but a few of the causes of hypopituitarism. Patients with hypopituitarism have an increased mortality rate.
https://www.sciencedirect.com/science/article/pii/B9780128041697000106

Physiologic and Pathophysiologic Alterations of the ... 翻译此页

Author: Kristin D. Helm, Ralf M. Nass, William … Publish Year: 2009

Hyperprolactinemia is a common cause of reproductive dysfunction in premenopausal women and is

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 49852

Manuscript Type: CASE REPORT

Recognizable type of pituitary, heart, kidney and skeletal dysplasia mostly caused by *SEMA3A* mutation: A case report

Fang Hu, Liao Sun

Abstract

BACKGROUND

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Recognizable type of pituitary, heart, kidney and skeletal dysp

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Holoprosencephaly and cleidocranial dysplasia in a patient ...

https://www.researchgate.net/publication/7618437_Holoprosencephaly_and_cleidocranial...

Holoprosencephaly **and** cleidocranial **dysplasia** in a patient due to two position-effect mutations: **Case report** and review of the literature Article · Literature Review in Clinical Genetics 68(4 ...

Published in: **Clinical Genetics** · 2005

Authors: Bridget A Fernandez · J Siegelbartelt · Joanne Herbrick · Ikuko Teshima · Stephen W ...

Affiliation: Memorial University of Newfoundland · Kaiser Permanente

About: Cartography · Chromosomal translocation · Position effect · Sonic hedgehog · Osteoc...

Clinical genetics Flashcards | Quizlet

https://**quizlet.com**/21239618/**clinical-genetics**-flash-cards

The fingers are **most** often affected but all joints can be involved. The joints may dislocate but can be popped back painlessly. Bruising and hematomas may arise after trivial injuries. Internal collagen defects: **heart** murmur (mitral valve prolapse) and weakened walls of ...

DAVID: Database for Annotation, Visualization, and ...

https://david.ncifcrf.gov/gene**Report**Full.jsp?rowids=1101,3696,3690,3691,3688,3694,3695...

Endoplasmic reticulum stress-mediated apoptosis contributes to a **skeletal dysplasia resembling platyspondylic lethal skeletal dysplasia, Torrance type**, in this line., **Serum** CTX-II levels in human brucellosis were higher than those of healthy controls but serum CTX-II levels in male patients were significantly higher than those of female patients indicating biological changes in cartilage and bone in ...

A differential diagnosis of inherited endocrine tumors and ...

https://**www.ncbi.nlm.nih.gov**/pmc/articles/PMC3715026

This syndrome is a tumor inherited condition that presents **PHEO/paragangliomas** in 5% of cases and in up to 13% of autopsies. NF1 is usually diagnosed at early ages in response to the presence of **neurofibromas**, **café-au-lait skin spots**, **skinfold freckling**, **iris Lisch nodules**, optic pathway gliomas, and bone dysplasia.

Cited by: 14 **Author:** Sergio P. A. Toledo, Delmar M. Lourenço,...

Publish Year: 2013