

## Match Overview

1	Internet 58 words crawled on 12-Oct-2015 <a href="http://www.ncbi.nlm.nih.gov">www.ncbi.nlm.nih.gov</a>	3%
2	Crossref 36 words Tomoko Lee, Yasuhiro Takeshima, Yo Okizuka, Kiyoshi H... mahira et al. "A Japanese child with geleophysic dysplasia c	2%
3	Internet 26 words crawled on 27-Jan-2020 <a href="http://www.nature.com">www.nature.com</a>	2%

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 62415

**Manuscript Type:** CASE REPORT

**Geleophysic dysplasia caused by a mutation in *FBN1*: A case report**

Ying Tao, Qing Wei, Xun Chen, Guang-Min Nong

### Abstract

#### BACKGROUND

Geleophysic dysplasia (GD) present the characterized clinical manifestations of acromelic dysplasia, including extremely short stature, short limbs, small hands and



Geleophysic dysplasia caused by a mutation in FBN1: case report an



ALL

IMAGES

VIDEOS

3,920 Results

Any time ▾

### [PDF] A Child with Geleophysic Dysplasia Type 2 Caused by a ...

<https://www.researchgate.net/publication/326898580...>

Objective: To report a geleophysic dysplasia type 2 (GD2) caused by FBN1 gene mutation, provide the clinical features of early diagnosis in patients with GD. Method Analysis was performed on clinical

### (PDF) A Child with Geleophysic Dysplasia Type 2 Caused by ...

<https://www.researchgate.net/publication/326898580...>

PDF | On Jan 1, 2018, 阳李 published A Child with Geleophysic Dysplasia Type 2 Caused by a Novel Mutation of FBN1: A Case Report and Literature Review | Find, read and cite all the research you ...

### The Clinical Cases of Geleophysic Dysplasia: One Gene ...

[https://www.hindawi.com/journals/crie/2018/8212417 ▾](https://www.hindawi.com/journals/crie/2018/8212417)

Geleophysic dysplasia is a rare multisystem disorder that principally affects the bones, joints, heart, and skin. This condition is inherited either in an autosomal dominant pattern due to FBN1 mutations or in an autosomal recessive pattern due to ADAMTSL2 mutations.

Cited by: 3

Author: Evgenia Globa, Nataliya Zelinska, Andrew...

Publish Year: 2018

### A Japanese child with geleophysic dysplasia caused by a ...

<https://www.researchgate.net/publication/232812652...>

Several years ago, heterozygous mutations in the TGF beta 5 region of the fibrillin-1 (FBN1) gene were found to cause both geleophysic dysplasia 2 (GD2) and AD, but with a highly variable clinical...

### OMIM Entry - # 231050 - GELEOPHYSIC DYSPLASIA 1; ...

[https://www.omim.org/entry/231050 ▾](https://www.omim.org/entry/231050)

Mar 26, 2019 · Geleophysic dysplasia-2 (GPHYSD2; 614185) is an autosomal dominant form of the disorder caused by heterozygous mutation in the FBN1 gene (134797) on chromosome 15q21.1.

### Three cases of Japanese acromicric/geleophysic dysplasia ...

<https://www.researchgate.net/publication/311553215...>

Three cases of Japanese acromicric/geleophysic dysplasia with FBN1 mutations: A comparison of clinical and radiological features January 2017 Journal of pediatric endocrinology & metabolism: JPEM ...





ALL

IMAGES

VIDEOS

11,900 Results

Any time ▼

## A chinese boy with geleophytic dysplasia caused by ...

<https://pubmed.ncbi.nlm.nih.gov/28917829>

**Geleophytic dysplasia**, belonging to the group of acromelic dysplasia, is a rare genetic disease. Two genes, FBN1 and ADAMTSL2, were known to be linked to this disorder. The disorder presents as extreme short stature, short limbs, small hands and feet, stubby fingers and toes, joint stiffness, toe walking, skin thickening, progressive cardiac valvular thickening and characteristic facial features, including a round ...

Cited by: 4

Author: Dongxiao Li, Hui Dong, Hong Zheng, Jinqin...

Publish Year: 2017

## A Japanese child with geleophytic dysplasia caused by a ...

<https://www.sciencedirect.com/science/article/pii/S0378111912013418>

Jan 10, 2013 · GD is a very rare disorder, and only 19 **cases** with **geleophytic dysplasia caused by mutations in FBN1** have been **reported** to date. In Japan, three **cases** of GD have been **reported**, including one **caused by a mutation** in ADAMTSL2 and one by a **mutation in FBN1** ; however, the detailed clinical course of this disease was not reported ( Le Goff et al., 2008 , Le Goff et al., 2011 , Matsui et al., 2002 ).

Cited by: 8

Author: Tomoko Lee, Yasuhiro Takeshima, Yo Okiz...

Publish Year: 2013

## A chinese boy with geleophytic dysplasia caused by ...

<https://www.sciencedirect.com/science/article/pii/S1769721217300848>

Dec 01, 2017 · Only two **cases** with **geleophytic dysplasia** type 2 due to **FBN1 mutations** have been reported (Wang et al. 2014). Here we report the first Chinese case of **geleophytic dysplasia** type 1 with a



 ALL
  IMAGES
  VIDEOS
  MAPS
  NEWS
  SHOPPING

11,900 Results

Any time ▾

## [A chinese boy with geleophysic dysplasia caused by ...](#)

<https://pubmed.ncbi.nlm.nih.gov/28917829>

**Geleophysic dysplasia**, belonging to the group of acromelic dysplasia, is a rare genetic disease. Two genes, FBN1 and ADAMTSL2, were known to be linked to this disorder. The disorder presents as extreme short stature, short limbs, small hands and feet, stubby fingers and toes, joint stiffness, toe walking, skin thickening, progressive cardiac valvular thickening and characteristic facial features, including a round ...

**Cited by:** 4

**Author:** Dongxiao Li, Hui Dong, Hong Zheng, Jinqin...

**Publish Year:** 2017

## [A Japanese child with geleophysic dysplasia caused by a ...](#)

<https://www.sciencedirect.com/science/article/pii/S0378111912013418>

Jan 10, 2013 · GD is a very rare disorder, and only 19 **cases** with **geleophysic dysplasia caused by mutations in FBN1** have been **reported** to date. In Japan, three **cases** of **GD** have been **reported**, including one **caused by a mutation** in ADAMTSL2 and one by **a mutation in FBN1** ; however, the detailed clinical course of this disease was not reported ( Le Goff et al., 2008 , Le Goff et al., 2011 , Matsui et al., 2002 ).

**Cited by:** 8

**Author:** Tomoko Lee, Yasuhiro Takeshima, Yo Okiz...

**Publish Year:** 2013

## [Three cases of Japanese acromicric/geleophysic dysplasia ...](#)

<https://pubmed.ncbi.nlm.nih.gov/27935852>

The genetic **cause** of AD and some **cases** of GD was shown to be **mutations** in the transforming growth factor (TGF)  $\beta$ -binding protein-like domain 5 of the fibrillin 1 gene (FBN1), which is also **mutated** in Marfan syndrome. In the present study, we **report** and compare the highly varied clinical and radiological features of three Japanese AD/GD children.

**Cited by:** 4

**Author:** Kosei Hasegawa, Chikahiko Numakura, Hir...

**Publish Year:** 2017

## [A chinese boy with geleophysic dysplasia caused by ...](#)

<https://www.sciencedirect.com/science/article/pii/S1769721217300848>

Dec 01, 2017 · Only two **cases** with **geleophysic dysplasia** type 2 due to **FBN1 mutations** have been reported (Wang et al., 2014). Here, we **report** the first Chinese **case** of **geleophysic dysplasia** type 1 with a missense **mutation** and a novel splicing **mutation** in ADAMTSL2. 2. Patient, materials and methods

**Cited by:** 4

**Author:** Dongxiao Li, Hui Dong, Hong Zheng, Jinqin...