

# World Journal of *Clinical Cases*

*World J Clin Cases* 2021 August 26; 9(24): 6964-7291



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**INDEXING/ABSTRACTING**

The *WJCC* is now indexed in Science Citation Index Expanded (also known as SciSearch®), Journal Citation Reports/Science Edition, Scopus, PubMed, and PubMed Central. The 2021 Edition of Journal Citation Reports® cites the 2020 impact factor (IF) for *WJCC* as 1.337; IF without journal self cites: 1.301; 5-year IF: 1.742; Journal Citation Indicator: 0.33; Ranking: 119 among 169 journals in medicine, general and internal; and Quartile category: Q3. The *WJCC*'s CiteScore for 2020 is 0.8 and Scopus CiteScore rank 2020: General Medicine is 493/793.

**RESPONSIBLE EDITORS FOR THIS ISSUE**

Production Editor: *Ji-Hong Lin*, Production Department Director: *Yn-Jie Ma*, Editorial Office Director: *Jin-Lei Wang*.

**NAME OF JOURNAL**

*World Journal of Clinical Cases*

**ISSN**

ISSN 2307-8960 (online)

**LAUNCH DATE**

April 16, 2013

**FREQUENCY**

Thrice Monthly

**EDITORS-IN-CHIEF**

Dennis A Bloomfield, Sandro Vento, Bao-Gan Peng

**EDITORIAL BOARD MEMBERS**

<https://www.wjgnet.com/2307-8960/editorialboard.htm>

**PUBLICATION DATE**

August 26, 2021

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<https://www.wjgnet.com/bpg/gerinfo/240>

**PUBLICATION ETHICS**

<https://www.wjgnet.com/bpg/GerInfo/288>

**PUBLICATION MISCONDUCT**

<https://www.wjgnet.com/bpg/gerinfo/208>

**ARTICLE PROCESSING CHARGE**

<https://www.wjgnet.com/bpg/gerinfo/242>

**STEPS FOR SUBMITTING MANUSCRIPTS**

<https://www.wjgnet.com/bpg/GerInfo/239>

**ONLINE SUBMISSION**

<https://www.f6publishing.com>

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

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**Author contributions:** Tao Y, Wei Q, and Nong GM conceptualized and designed the study; Tao Y and Wei Q drafted the initial manuscript; Chen X collected and analyzed the data; Tao Y, Wei Q, and Chen X reviewed and revised the manuscript; Nong GM coordinated and supervised the data collection, and critically reviewed the manuscript; All authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

**Informed consent statement:**

Informed written consent was obtained from the patients for publication of this report and any accompanying images.

**Conflict-of-interest statement:** The authors have no potential conflicts of interest to disclose.

**CARE Checklist (2016) statement:**

The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

**Open-Access:** This article is an open-access article that was selected by an in-house editor and

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### Abstract

#### BACKGROUND

Geleophysic dysplasia (GD) presents the characterized clinical manifestations of acromelic dysplasia, including extremely short stature, short limbs, small hands and feet, stubby fingers and toes, joint stiffness and others. It is clinically distinct from the other acromelic dysplasia in terms of symptoms such as cardiac valvular abnormalities, progressive hepatomegaly and tracheal stenosis.

#### CASE SUMMARY

We report on a Chinese 9-year-old girl with GD with the c.5243G>T (p.C1748F) mutation in *FBN1* (fibrillin 1, OMIM 134797). She was born in Guangxi Zhuang Autonomous Region of China. The patient presented with typical clinical features of GD and recurrent respiratory tract infections over 6 years. Laboratory studies and chest computed tomography (CT) scan indicated bronchopneumonia. Her echocardiography revealed mild mitral valve thickening with regurgitation. Laryngopharyngeal CT and electronic bronchoscopy revealed severe glottic stenosis. Echocardiography examination displayed mild mitral valve thickening and regurgitation. Ophthalmic examination did not reveal myopia or lens dislocation. Treated with ceftriaxone sodium and methylprednisolone sodium succinate for injection as well as methylprednisolone orally, patient's symptoms had improved.

#### CONCLUSION

GD is a rare genetic condition that can cause life-threatening cardiovascular and respiratory problems. This study also found that the identified genotype of GD could be related to different clinical phenotypes.

**Key Words:** Fibrillin 1; Geleophysic dysplasia; Acromelic dysplasia; Short stature; Tracheal stenosis; Case report

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**Manuscript source:** Unsolicited manuscript

**Specialty type:** Pediatrics

**Country/Territory of origin:** China

**Peer-review report's scientific quality classification**

Grade A (Excellent): 0  
Grade B (Very good): B  
Grade C (Good): C  
Grade D (Fair): 0  
Grade E (Poor): 0

**Received:** February 23, 2021

**Peer-review started:** February 23, 2021

**First decision:** May 11, 2021

**Revised:** May 19, 2021

**Accepted:** July 7, 2021

**Article in press:** July 7, 2021

**Published online:** August 26, 2021

**P-Reviewer:** Malik S, Zavras N

**S-Editor:** Liu M

**L-Editor:** Filipodia

**P-Editor:** Yuan YY



**Core Tip:** We aim to report a 9-year-old girl with geleophysic dysplasia (GD) with mutation c.5243G>T (p.C1748F) in *FBN1*. Other than the patient we reported, a total of 9 acromelic dysplasia cases due to mutations in c.5242T, c.5243G or c.5244T of *FBN1* have been reported, which all are predicted to result in the substitution of cysteine at codon 1748. This study also found that the identified genotype of GD could be related to different clinical phenotypes.

**Citation:** Tao Y, Wei Q, Chen X, Nong GM. Geleophysic dysplasia caused by a mutation in *FBN1*: A case report. *World J Clin Cases* 2021; 9(24): 7175-7180

**URL:** <https://www.wjgnet.com/2307-8960/full/v9/i24/7175.htm>

**DOI:** <https://dx.doi.org/10.12998/wjcc.v9.i24.7175>

## INTRODUCTION

Geleophysic dysplasia (GD) belongs to acromelic dysplasia and has unique features [1]. GD is clinically distinct from other acromelic dysplasia in its symptoms such as cardiac valvular abnormalities, progressive hepatomegaly and tracheal stenosis [1]. Three genes, *ADAMTSL2* (OMIM 612277), *FBN1* and *LTBP2* (OMIM 602091), have been associated with GD [2-4]. Nearly all the mutations in *FBN1* associated with GD are located in exons 41 and 42 [5]. *FBN1*, located at 15q21, encodes an extracellular matrix protein that forms a major component of microfibrils of the extracellular matrix in connective tissues [6]. The most common inherited disease caused by *FBN1* mutations is Marfan syndrome. In contrast to the bone overgrowth and tall stature in individuals with Marfan syndrome, heterozygous mutations in the transforming growth factor (TGF)- $\beta$ -binding protein-like domain 5 (TB5) of *FBN1* have been identified as the underlying etiology of acromelic dysplasia, including GD, which manifest as heritable short stature syndromes [7]. Both Marfan syndrome and *FBN1*-related acromelic dysplasia have autosomal dominant inheritance [6,8]. We reviewed the literature of reported acromelic dysplasia cases due to mutations at c.5242T, c.5243G and c.5244T of *FBN1* by searching PubMed from 2000 onwards.

## CASE PRESENTATION

### Chief complaints

A 9-year-old Chinese girl presented with labored breathing, cough with wheeze and nasal discharge.

### History of present illness

The patient had no intellectual development disorder, hepatomegaly or hand joint stiffness.

### History of past illness

The patient had a history of hypothyroidism, short stature, obstructive sleep apnea hypopnea syndrome and recurrent respiratory tract infections (RRTIs).

### Personal and family history

The patient was born in Guangxi Zhuang Autonomous Region of China and had no special personal or family history.

### Physical examination

The patient had extremely short stature (94 cm, < -3 SD) (Figure 1A), low weight (17 kg, < -3 SD) [9], unique facial features (round face, small nose with anteverted nostrils, broad and depressed nasal bridge and thin upper lip) (Figure 1B), short limbs, and short hands and feet (Figure 1C and D).

### Laboratory examinations

Laboratory tests suggested inflammation.



**Figure 1 Clinical characters of the patient.** A, C and D: Patient presented extreme short stature, short limbs, short hands and feet; B: Patient had unique facial features including round face, small nose with anteverted nostrils, broad and depressed nasal bridge and thin upper lip.

### **Imaging examinations**

Her chest computed tomography (CT) scan indicated bronchopneumonia. Laryngopharyngeal CT revealed tracheal stenosis (Figure 2A and B).

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## **FINAL DIAGNOSIS**

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GD.

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## **TREATMENT**

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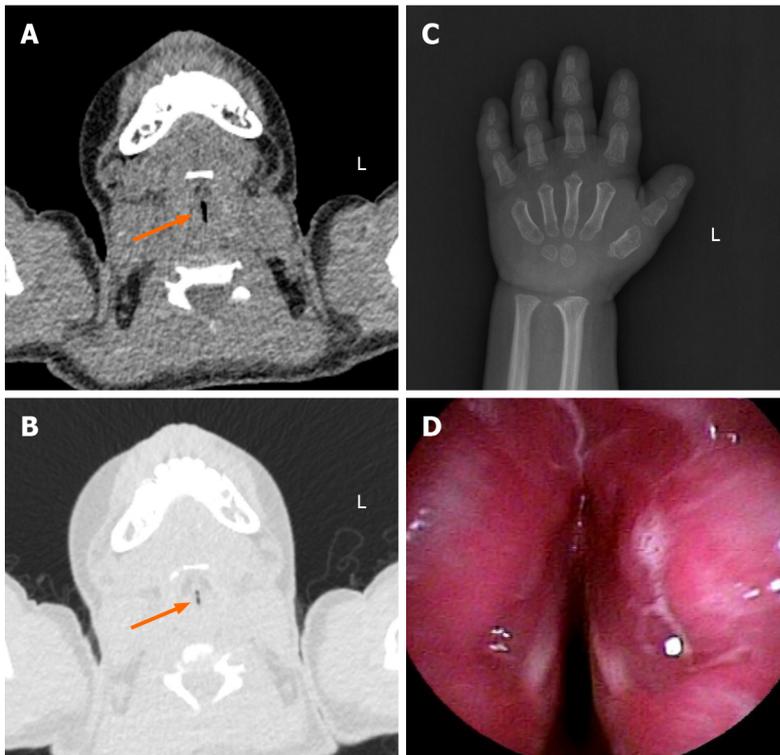
The patient presented an oxygen saturation of 96% with a nasal catheter giving 0.5-1 L/min oxygen inhalation. She received human immunoglobulin (50 mL: 2.5 g [5%]) intravenously on the second and the last day of admission. Isopropyl compound ipratropium bromide solution (inhalation of Combivent aerosol budesonide) 1.25 mL + (Pulmicort) 200 µg + normal saline 1 mL atomization inhalation were continued daily during the hospital stay. The patient received 500 mg/d ceftriaxone sodium for injection until day 11, when the symptoms of the infection were resolved. For antiasthma management, the patient received methylprednisolone sodium succinate for injection at 40 mg/d up to day 10 and methylprednisolone at 16 mg/d orally daily until discharge. The patient was discharged on day 13 after progressive improvement in respiratory function.

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## **OUTCOME AND FOLLOW-UP**

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When discharged, patient's symptoms of RRTIs had improved by using methylprednisolone 20 mg/d orally. Patient returned to the outpatient department regularly.



**Figure 2 Representative radiographic and flexible bronchofiberscope images.** A and B: Laryngopharynx computed tomography presented trachea stenosis; C: Skeletal X-ray was taken at age 6 and revealed a delayed bone age and epiphyseal dysplasia; D: Electronic bronchoscope demonstrated severe glottic stenosis.

## DISCUSSION

GD, acromicric dysplasia (AD) and Weill-Marchesani syndrome (WMS) are kinds of acromelic dysplasia[1], but these three disorders have their own unique features. GD is clinically distinct from AD and WMS in its symptoms such as cardiac valvular abnormalities, progressive hepatomegaly and tracheal stenosis. Ordinarily, conditions that resemble GD but without cardiac valvular abnormality are diagnosed as AD, and those accompanied by distinguishing eye anomalies, including lenticular myopia, ectopia lentis, glaucoma and spherophakia, are diagnosed as WMS[1].

Like the patients who have been reported to have *FBN1*-related diseases, the present patient also displayed an autosomal dominant inheritance pattern and had a mutation in the TB5 domain of *FBN1*. The inheritance method of *FBN1* shows autosomal dominance, whereas the inheritance of *ADAMTS10*, *ADAMTS17* and *LTBP2* is autosomal recessive[3]. Mutations in these genes are reported to cause WMS[10], which has similar clinical manifestations as GD. *ADAMTS10*, *ADAMTS17* and *LTBP2* were all included in our genetic analysis, and none of them had mutations.

We reviewed the literature of reported acromelic dysplasia cases, including GD, AD and WMS, due to mutations at c.5242T, c.5243G and c.5244T of *FBN1*, which all are predicted to result in the substitution of cysteine at codon 1748. Other than the patient we reported, a total of 9 patients were found, including one family with 7 patients. Patient 1[2] had the heterozygous mutation c.5243G>C (p.Cys1748Ser). Patient 2[1], like the girl we reported, had the same heterozygous mutation c.5243G>T (p.C1748F). Patient 3[11], the proband of the family, had the heterozygous mutation c.5242T>C (p.C1748R). There was no mutation reported at position c.5244T. All the patients had progressive growth delays from an early age and presented dysmorphic features such as short stature, short limbs and stubby fingers and toes. No severe abnormalities were mentioned in either the mitral or tricuspid valve. However, Patient 3[11] developed a life-threatening subacute aortic dissection extending from the aortic root to the left subclavian artery of the thoracic aortic arch. Severe tracheal stenosis developed in our patient and Patient 1[2]. Thyroid hypofunction was found in both our patient and Patient 2[1]. It is worth noting that Patient 2, who had the same mutation as our patient, was diagnosed with WWS and had small, round lenses and moderate myopia but did not develop tracheal stenosis (Table 1).

**Table 1 Clinical and genetic characteristics of acromelic dysplasia cases due to mutations in c5242T, c5243G of *FBN1* (+: Present, -: Absent, NA: Not available)[1,2,11]**

	Our patient	Patient 1[2]	Patient 2[1]	Family[11]			
				Patient 3 (proband)	Proband's father	Proband's sister	Proband's children
Mutation	c.5243G>T (p.C1748F)	c.5242T>C (p.Cys1748Ser)	c.5243G>T (p.C1748F)	c.5242T>C (p.C1748R)			
Disorder	GD	GD	WMS	WMS			
Short stature	+	+	+	+	+	+	+
Short limbs	+	+	+	+	+	+	+
Stubby fingers and toes	+	+	+	+	+	+	+
Mitral valve thickening and regurgitation	Mild	-	NA	-	NA	NA	NA
Aortic dissection	-	-	-	+	-	-	-
Tracheal stenosis	+	+	-	-	-	-	-
Myopia	-	+	+	+	+	+	NA
Ectopia lentis	-	+	+	+	+	+	NA
Thyroid hypofunction	+	-	+	NA	NA	NA	NA
Hepatosplenomegaly	-	-	-	NA	NA	NA	NA

NA: Not available; GD: Geleophysic dysplasia; WMS: Weill-Marchesani syndrome.

The present patient was diagnosed with GD due to the presence of mitral valve abnormalities and tracheal stenosis. However, Patient 2[1], with the same mutation as our patient, was diagnosed with WMS and lacked either mitral valve abnormalities or tracheal stenosis. These findings demonstrate that an identified genotype can be related to different clinical phenotypes. In addition to genotypes, environmental factors also play an important role in phenotype development. In this study, both the patient we reported and Patient 1[2] developed persistent tracheal stenosis with age. It is noteworthy that both of them had a history of RRTIs at an early age before having developed persistent tracheal stenosis. However, none of the same conditions were mentioned in Patient 2[1]. RRTIs might play an important role in tracheal stenosis development at an early age.

Progressive cardiac valvular abnormality is a common cause of death in patients with GD[12,13]. In this study, no severe abnormality was found in the mitral or tricuspid valve. However, Patient 3 developed a life-threatening subacute aortic dissection extending from the aortic root to the left subclavian artery of the thoracic aortic arch, which might be caused by aortic valve abnormalities. Moreover, severe tracheal stenosis developed in our patient and Patient 1, the latter of whom needed tracheostomy permanently. This demonstrated that respiratory problems, especially tracheal stenosis, might also be the cause of death in patients with GD. We should pay attention to cardiovascular and respiratory problems in patients with GD to prevent a severe, even life-threatening, event from occurring and to treat complications as early as possible.

Other than Patient 2[1], none of the patients with acromelic dysplasia were reported to present thyroid hypofunction. Our patient had mild thyroid hypofunction without impaired intelligence. The patient took Euthyrox orally and regularly, but her growth delay showed no improvement. This result indicates that thyroid hypofunction is one of the accompanying manifestations, rather than a causative factor, of GD. Interestingly, both our patient and Patient 2[1] had the same mutation, c.5243G>T (p.C1748F) of *FBN1*, which demonstrates that thyroid hypofunction may be specific to patients with the c.5243G>T (p.C1748F) of *FBN1* in acromelic dysplasia.

## CONCLUSION

GD is a rare genetic condition that can cause life-threatening cardiovascular and respiratory problems. This study also found that the identified genotype of GD could be related to different clinical phenotypes.

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