

## Case Report

# Bilateral morgagni hernia in a case of Weill-Marchesani syndrome- a rare association

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### ABSTRACT

Morgagni hernia constitutes only about 2% of all diaphragmatic hernias and bilateral Morgagni hernia is extremely rare. Here we present a 75 year old female patient with morphometric features of Weill-Marchesani syndrome who has bilateral Morgagni hernia. This association is reported for the first time in literature.

**Keywords:** Foramen of Morgagni, Morgagni hernia, Wiell-Marchesani syndrome

## INTRODUCTION

Weill-Marchesani syndrome is due to connective tissue abnormality and is inherited as autosomal dominant or recessive disorder. Clinical features of this syndrome include short stature, stiffness of joints of upper limbs, microspherophakia, *ectopia lentis*, low set flat palate and other bony abnormalities. Since connective tissue is defective in this condition weakness of fibro-muscular structures like diaphragm may be expected. Morgagni hernia occurs through the anteriolateral foramen of diaphragm and is mostly seen on the right side. Bilateral Morgagni hernia is extremely rare. It is proposed that weakness of the diaphragmatic muscle fibers may be the cause for this rare presentation in Weill-Marchesani syndrome.

## CASE REPORT

A 75 year old female patient presented to the Pulmonary Medicine outpatient department. She was being referred by her treating physician as a case of right lower lobe pneumonia. She was a diabetic and hypertensive for the last 8 years and was well controlled on medication. She used to work as a head load worker in a coffee plantation

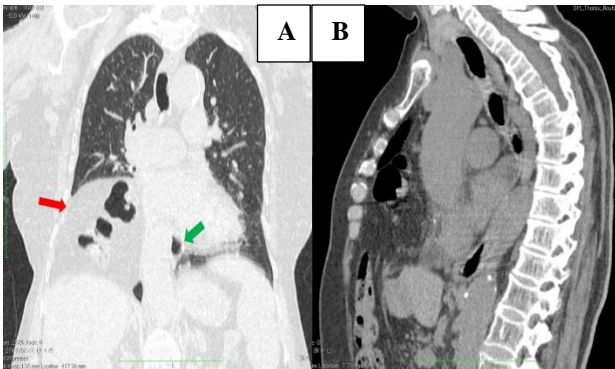
and is leading a retired life at present. She was never a smoker but used to chew tobacco. She had six live births and gives history of four term babies dying in utero.



**Figure 1: A) X-ray chest PA view showing nonhomogeneous opacity right lower zone; B) X-ray of left elbow demonstrating fixed flexion deformity of the elbow joint.**

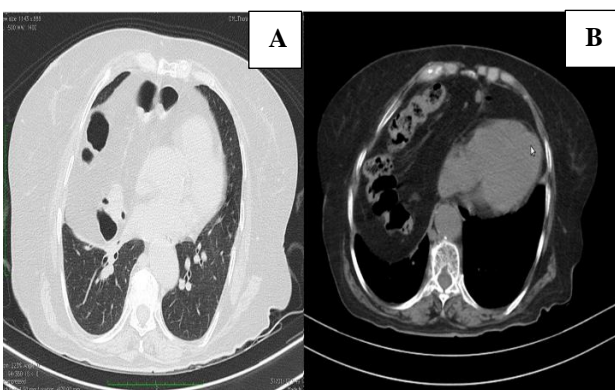
On examination she was of short stature and poorly nourished. She had a height of 138cm and weight of 34kg with a BMI of 17.8. She has stiffness of both elbow joint

with restricted extension (Figure 1B). She had a shallow palatal arch. There was exaggerated thoracic kyphosis and scoliosis to the left. Chest movements were diminished on right lower part with impaired percussion note. Breath sound was diminished on right side and coarse crackles are heard on right lower part. Ophthalmologic examination does not reveal any abnormal findings. Based on short stature, stiff elbow joints, low set palatal arch and kyphoscoliosis she was diagnosed to have Weill Marchesani syndrome which is a genetic disorder with defect in connective tissue.



**Figure 2: CT Thorax sagittal view demonstrating hernia through the foramen of Morgagni; A) Block arrows showing hernia on either side.**

A presumptive diagnosis of right lower lobe pneumonia with sympleuronic effusion was made. Blood routine and sputum examinations were within normal limits. Her symptoms responded to antibiotic and symptomatic measures. The follow up X-ray showed a non-homogenous opacity in the right lower zone (Figure 1A). CT thorax revealed that the shadow was due to herniation of abdominal contents to thorax (Figure 2). It demonstrated bilateral Morgagni hernia (Figure 3A). There was omentum and transverse colon herniating through right foramen and omentum herniating through left foramen with a definite cleavage separating the two (Figure 3B).



**Figure 3: A) CT Thorax axial window showing the hernia; B) demonstrates contrast filled transverse colon herniating on the right side. Omental herniation on the left side also is visible.**

## DISCUSSION

Weill-Marchesani syndrome (WMS) is a disorder of connective tissue. As connective tissue forms the body's supportive framework, providing structure and strength to the muscles, joints, organs, and skin; this syndrome has unique abnormalities in different organ systems. The major signs and symptoms of WMS include short stature, eye abnormalities, unusually short fingers and toes (brachydactyly), and joint stiffness especially in upper limbs. The eye abnormalities include small, sphere-shaped lens (microspherophakia) and ectopia lentis.<sup>1</sup> These patients may later develop glaucoma, leading to blindness. Occasionally heart defects or an abnormal heart rhythm can occur in people with WMS.

Weill-Marchesani syndrome appears to be rare with an estimated prevalence of 1 in 100,000 people. WMS can be inherited as an autosomal recessive or an autosomal dominant pattern. Mutations in the ADAMTS10 and FBN1 genes can cause WMS syndrome. The ADAMTS10 gene provides instructions for making a protein which is important for normal growth before and after birth, and it appears to be involved in the development of the eyes, heart, and skeleton. Mutations in this gene disrupt the normal development of these structures, which leads to the specific features of WMS. The FBN1 gene provides instructions for making a protein called fibrillin-1. This protein is needed to form micro fibrils that provide strength and flexibility to connective tissue. The FBN1 mutation responsible for WMS leads to an unstable version of fibrillin-1. This unstable protein interferes with the normal assembly of micro fibrils, which weakens connective tissue and causes the abnormalities associated with WMS. This can affect all fibromuscular structures including diaphragm.

Homozygous mutations in the ADAMTS10 gene cause Weill-Marchesani syndrome 1 (WMS 1). Weill-Marchesani syndrome 2 (WMS 2) is a clinically similar syndrome but results from heterozygous mutations in FBN1. Homozygous mutations in ADAMTS17 cause the Weill-Marchesani-Like syndrome.<sup>2</sup> It is not always possible to distinguish between the AR and AD forms of the disease using clinical criteria alone.

Congenital Morgagni Hernia (CMH) is rare, constituting roughly 2% of all congenital diaphragmatic hernias (CDH).<sup>3</sup> Herniation of abdominal content occurs through anterolateral or triangular parasternal gaps in the diaphragm.<sup>4</sup> This mostly occurs on the right side, though they can rarely occur on the left or bilaterally.<sup>5</sup> They are generally asymptomatic and incidentally found during an unrelated diagnostic workup.<sup>6</sup> Berman et al treated only 18 cases of CMH over a period of 40 years and Pokorny et al reported only 4 cases of CMH over a period of 25 years.<sup>8,9</sup> Cigdem et al treated 16 cases of CMH over a period of 23 years.<sup>9</sup> Giovanni Battista Morgagni, an Italian anatomist, first described the herniation of abdominal contents through the sternochondral triangles

in 1769 based on cadaver observation.<sup>10</sup> Later, in 1828, Larrey described a surgical approach to the pericardial sac through these same triangles. The costochondral triangles have been referred to as the foramen of Morgagni on the right and the space of Larrey on the left. They form when the pars sternalis and a costochondral arch fuse and close around the internal thoracic artery as it becomes the superior epigastric artery. Occasionally these spaces do not fully close and allow for the herniation of abdominal contents into the thorax.<sup>4,11</sup>

Morgagni hernias can be diagnosed during any period of life including prenatal period.<sup>12</sup> About 90% of these hernias occur on the right side of the sternum and 8% on the left.<sup>13</sup> Bilateral hernia makes up 2% of all Morgagni hernias. The hernias most frequently contain omental fat and transverse colon. They rarely contain liver or stomach.<sup>14</sup> Sometimes herniation occurs into the pericardial cavity. When this occurs, serious cardiorespiratory compromise may result.<sup>15,16</sup> In adults, Morgagni hernia is also associated with obesity, trauma, weight lifting, or other causes of increased intra-abdominal pressure. Our patient was a head load worker and this can be a predisposing factor. In the pediatric age group, the presentation of CMH is variable and non-specific. Usually the presentation is that of recurrent chest infection and rarely may present with gastrointestinal symptoms. Our patient also reported with respiratory symptoms rather than gastrointestinal symptoms. Majority of patients suffered repeated attacks of chest infections and received several courses of antibiotics.

There is an increased risk of associated anomalies with CMH, with a variable incidence ranging from 34% to 50%.<sup>5</sup> One study reported as high as 78.3% associated anomalies in CMH including 34.8% congenital heart disease.<sup>6</sup> ASD and VSD were the commonest associated heart defects. This however did not influence the final outcome and had no effect on the overall morbidity. An interesting association was that of CMH and Down's syndrome. In a collective series of 46 children with Morgagni hernia, 16 (34.8%) of them had Down's syndrome.<sup>17</sup> Cigden et al in a series of 16 patients with CMH seen over a period of 23 years reported a 31.25% incidence of Down's syndrome.<sup>9</sup> Other rare clinical associations of Morgagni hernia are pulmonary hypoplasia, gastric volvulus, rotational abnormalities and midgut volvulus, hypoplasia of the left ventricle and bilateral renal hypertrophy.

Diagnosis of diaphragmatic hernias is easily made with chest radiographs, ultrasound (US), or computed tomography (CT). Chest radiography often requires anterior-posterior images to evaluate hernia severity and lateral images to evaluate hernia location. Images can vary depending on the contents of the hernia. Solid viscera protruding through the hernia may show an opaque hemithorax with or without mediastinal shift. Hollow viscera are often present as loops of bowel within

the thorax. An anterior medial mass on chest radiography can be suggestive of a Morgagni hernia. Radiological differential diagnoses include pneumonia, atelectasis, diaphragmatic eventration, mediastinal lipoma, liposarcoma, abscess, and pleuropericardial cyst.<sup>18</sup>

Computed tomography can be used to confirm a suspected diaphragmatic hernia. Multiphase CT can demonstrate the diaphragm and the organs that herniate through it. Omental vessels can be visualized in some diaphragmatic hernias and can help differentiate it from lipomas or liposarcomas. Intravenous contrast can help enhance these vessels and confirm a diagnosis.<sup>14</sup> Ultrasound is helpful in assessing diaphragmatic hernias that contain solid viscera. Hepatic echo-texture and color Doppler sonogram can confirm liver in thorax.<sup>19</sup> Hernias that contain hollow viscera can be more difficult to evaluate with US. Air in the bowels and lungs, as well as rib shadowing, can often distort the images and make them difficult to interpret.<sup>14</sup>

The association of Weill Marchesani syndrome and bilateral Morgagni hernia is not previously reported in literature. Defective collagen in Weill Marchesani syndrome may be the cause for persistence of defect in the diaphragm leading to herniation. Thus causative relationship between the two clinical entities can be postulated. We propose that this is an extended syndrome of Weill Marchesani syndrome and would like to name it as Weill-Marchesani- Wayanad syndrome giving due credit to the place from where it is first reported.

## CONCLUSION

Weill-Marchesani syndrome is an inherited disorder in which there is defect in the collagen tissue. This can present with different manifestations in the body. Here we present a case of Weill-Marchesani syndrome with bilateral Morgagni hernia and postulate that defective collagen may be the pathogenetic mechanism for persistence of foramen of Morgagni leading to herniation later. This association is being reported for the first time in literature.

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