CASE REPORT

Plummer-Vinson syndrome with pectus carinatum mitral valve prolapsus and exotropia in an 18-year-old boy

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SUMMARY

The Plummer-Vinson syndrome is a clinical syndrome characterised by dysphagia, web or webs in upper oesophagus and iron-deficiency anaemia. The syndrome is often seen in women of age 40–70 years and rarely in adolescents. Plummer-Vinson syndrome might be associated with malignancy, myeloproliferative disorder and autoimmune diseases including coeliac disease, rheumatoid arthritis and Sjögren syndrome. However, according to our literature search, there are no reports of such case associated with thorax deformity, cardiac pathology and ocular findings. We present a case of an 18-year-old boy with a rare presentation of this syndrome including pectus carinatum, exotropia and mitral valve prolapsus.

CASE PRESENTATION

An 18-year-old boy presented to our clinic with dysphagia against solid food for approximately 1 year, which aggravated in the past 3 months, with fatigue, weakness, nausea, vomiting and weight loss. Dysphagia had been against solid food for 1 year and had become against solid as well as liquid food in the past 3 months. The patient defined a weight loss of 5 kg in the past 6 months due to dysphagia and vomiting. The patient had been receiving an irregular iron replacement therapy for iron-deficiency anaemia and had not received any medications in the past 4 years.

The physical examination revealed that the patient was pale with stable vital signs. Conjunctivas were also pale and the patient had strabismus and asymmetrical protruding on the right thorax wall. Cardiac examination revealed mid-systolic click and late systolic murmur. Other examination findings were normal.

Laboratory examination revealed that the level of haemoglobin was 9.7 (13.5–16) g/dL, haematocrit 32.2 (35–44)%, the mean corpuscular volume 65.4 (78–90) fL and the platelet count 469 (150–400) $\times 10^3/\mu\text{L}$. The serum iron level was 12 (45–182) $\mu\text{g/dL}$. Iron binding capacity was 401 (110–370) $\mu\text{g/dL}$, transferrin saturation 3% and ferritin level 1.9 (24–336) ng/mL. The biochemical parameters, thyroid function tests and autoantibody tests were within normal ranges, and examination of blood in faeces resulted negative.

Medical anamnesis revealed that strabismus had been present since infancy and an ocular examination revealed altered exotropia (figure 1A). An asymmetrical protrusion was observed on the thorax wall (figure 1B) and computed thorax tomography was performed to examine the thorax deformity. The thorax deformity was diagnosed as a chondrogladiolar type of asymmetrical pectus carinatum (figure 2B). Cardiac examination revealed a mid-systolic click and late systolic murmur; trans-thoracic echocardiography was therefore performed and mitral valve prolapsus was demonstrated (figure 2A). Esophagoscopy was performed to elucidate dysphagia and a web formation was observed in the upper oesophagus that blocked the passage of the scope (figure 3A,B). Dilation treatment was performed since the patient was symptomatic and iron replacement therapy was initiated.

DISCUSSION

The exact aetiopathogenesis of this syndrome and oesophageal webs is still unknown. Possible

BACKGROUND

The Plummer-Vinson syndrome (PVS) is a clinical syndrome often seen in adult women and characterised by dysphagia, web or webs in upper oesophagus and iron-deficiency anaemia. The disease is termed PVS in the USA and Paterson-Brown-Kelly syndrome in the UK.¹ It was first reported by Plummer in 1912 with 21 cases characterised by oesophageal diffuse dilation and spasm.¹ Vinson has defined another case of oesophageal angulation in 1919 and attributed this to the case previously described by Plummer.² The characteristic clinical findings of the syndrome were first defined by Paterson and Kelly in 1919.^{3–4} There are no exact data regarding the frequency of the syndrome. Although it had been more common among the Scandinavian population at the onset of the 20th century, this syndrome has now become rarer with an improved nutrition and effective treatment of iron-deficiency anaemia. Most of the patients are white middle-aged women, in the fourth to seventh decade of life; but some cases in children and adolescents have been reported.^{5–6} PVS might be associated with malignancy, myeloproliferative disorder and autoimmune diseases including coeliac disease, rheumatoid arthritis and Sjögren syndrome.^{7–9} However, according to our literature search, there are no reports of this case associated with thorax deformity, cardiac pathology and ocular findings. We present a case of an 18-year-old boy with a rare presentation of this syndrome including pectus carinatum, exotropia and mitral valve prolapsus.

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To cite: Sahin C, Ozseker B, Rencuzogullari I, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-202047

Figure 1 (A) Altered exotropia. (B) Asymmetrical pectus carinatum.

aetiological factors include genetic, nutritional and immunological reasons.⁵ Although reports of higher incidence among relatives suggest a hypothesis of familial predisposition, the most significant potential aetiological factor is believed to be iron-deficiency anaemia.⁵ The iron-deficiency theory assumes that the decrease in iron-dependent oxidative enzymes leads to atrophy of the pharynx muscles and mucosa, and thus web formation.¹⁰ Improvement in dysphagia with effective treatment of iron-deficiency anaemia and a decreased incidence of the syndrome with modern nutrition are supportive of this theory.¹⁰

PVS is thought to be associated with malignancy, myeloproliferative disorder and autoimmune diseases. The aetiological role of autoimmunity is suspected since PVS is often accompanied by autoimmune diseases such as coeliac disease, rheumatoid arthritis, Sjögren syndrome and autoimmune thyroid disease.¹¹ However, according to our literature search, there are no reports of this case associated with thorax deformity, cardiac pathology and ocular findings. We could not establish a relation

between PVS and these manifestations. Currently, there is a lack of knowledge to determine whether or not these manifestations are part of PVS. It is our recommendation that further investigations regarding patients with PVS be performed in order to determine a possible relation.

Most webs are asymptomatic and are observed in 5–15% of patients with dysphagia.¹² Oesophageal webs often consist of multiple layers of squamous epithelium and muscular mucosa.¹³ Webs might be multiple, and are often located in the upper oesophagus.¹³ Dysphagia in PVS is often painless and typically intermittent.¹³ Occasionally, it may aggravate progressively and present with weight loss due to blockage of oral intake.⁵ The oesophageal motility is also impaired in PVS. Oesophageal dysmotility, in addition to oesophageal webs, have been found to contribute dysphagia.¹⁰

PVS is associated with an increased risk of squamous cell carcinoma of the oesophagus.⁵ Development of oesophageal or pharyngeal carcinoma has been reported in 3–15% of patients, with PVS often consisting of women over 50 years of age.¹⁴ Carcinoma might present years after the onset of dysphagia. Therefore, cases should be followed closely with endoscopic and other imaging modalities for the hypopharynx and upper oesophagus. A surveillance upper gastrointestinal endoscopy is recommended every year.⁵

Oesophageal webs can be detected using barium swallow X-rays, videofluoroscopy and upper gastrointestinal endoscopy. Webs may sometimes be overlooked in endoscopy and may be ruptured accidentally. Endoscopic examination of the upper gastrointestinal system should be performed very carefully in suspected cases with webs.

PVS can be treated easily and effectively with iron replacement and mechanical dilation. In many patients, iron

Figure 2 (A) Transthoracic echocardiographic image of mitral valve prolapsus. (B) Chondrogladiolar type of asymmetrical pectus carinatum image in thorax tomography.

Figure 3 (A) and (B) endoscopic images of oesophageal web.

replacement can resolve dysphagia.⁵ Particularly, in elderly patients, the aetiology of iron deficiency should be elucidated to rule out an active haemorrhage or malignancy in every case of PVS. The anaemia improves with isolated iron replacement therapy in the majority of cases.¹⁵ The clinical studies have demonstrated that iron replacement therapy contributes to the improvement of dysphagia by improving oesophageal motility in these patients.¹⁰ Iron replacement should continue until haematocrit and ferritin levels return to normal. However, patients with a prolonged dysphagia and severe symptoms are often refractory to iron replacement therapy, and mechanical dilation of the stenosis is required.¹⁶ A single performance of mechanical dilation is often adequate to resolve dysphagia. There are a limited number of patients requiring surgical excision.¹⁶

Mechanical dilation was the first choice of treatment in our case since there was persistent dysphagia and severe symptoms leading to weight loss. Dysphagia improved rapidly following

dilation. Iron replacement therapy was initiated and the control haemogram level was 14.7 g/dL at 2 months. The patient's appetite was improved and he gained 5 kg in 2 months.

Contributors CS contributed in writing; BO in literature search; IR in concept and design; and AZ in supervision and data collection.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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Learning points

- This case suggests that a diagnosis of Plummer-Vinson syndrome (PVS) should be considered in patients with iron-deficiency anaemia and dysphagia.
- Such patients should be included in a screening programme with upper gastrointestinal endoscopy and other imaging modalities since PVS is a known risk factor in squamous cell carcinoma of the upper gastrointestinal system.
- In addition, patients with PVS should be screened for other associated pathologies.
- Further investigations should be performed in order to determine a possible relation between PVS and these pathologies.

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