CASE REPORT

Plummer Vinson syndrome in a male and his chromosomal study – A case report

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Received 20 March 2015; accepted 3 April 2015
Available online 8 May 2015

KEYWORDS
Plummer Vinson syndrome; Esophageal web; Post-cricoid carcinoma; Chromosomal aberration

Abstract  Plummer Vinson syndrome (PVS) is a triad of iron deficiency anemia, esophageal web and dysphagia. The exact etiology of PVS remains controversial but it has been associated with nutritional deficiency, autoimmune disorders, hereditary factors and remarkable high female predominance. This paper reports an atypical presentation of PVS in a 38 year old Indian male with special emphasis given on chromosomal analysis. Chromosomal assessment is done as it is a good predictor of the possibility of development of post-cricoid carcinoma (PCC) in patients with PVS. Chromosomal aberrations like translocation, gain, loss, breakpoints and duplications are studied and they revealed normal male chromosomal pairing.

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1. Introduction

Plummer–Vinson syndrome (PVS) is characterized by dysphagia which is usually due to upper esophageal web, anemia, splenomegaly and other features like angular cheilitis and glossitis [1]. It is also known as Paterson–Kelly syndrome or sideropenic anemia with epithelial lesions. This syndrome is extremely rare but it is important because it identifies the risk of squamous cell carcinoma of the postcricoid area and upper esophagus. Most of the patients are middle aged women, in the fourth to seventh decade of life but the syndrome has also been described in children and adolescents [2]. The dysphagia in PVS is usually painless and intermittent or progressive over years, limited to solid foods and sometimes associated with weight loss. Exact etiopathogenesis of PVS is not known. The incidence of upper aero-digestive tract carcinoma in PVS is between 4% and 16% and almost occurs in the postcricoid region [3]. The high turnover rate of the epithelium at the upper digestive tract makes the subject vulnerable to iron deficiency because of the deficiency of iron dependent enzymes. This reduction of oxidative enzymes of epithelial cells, free radicals stress and DNA damage may lead to mucosal atrophy, pharyngoesophageal ulcerations and esophageal webs.

Herein we present a case of PVS in adult male with long standing dysphagia and sideropenia. Keeping in mind genetic alterations which may occur during the development of

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Peer review under responsibility of Ain Shams University.
2. Case report

A 38 year old male presented to the outpatient department of Otorhinolaryngology with foreign body sensation in the throat since one year. He sometimes experienced choking sensation and aspiration during eating. On general examination, the patient’s vital signs were normal, and he had thin body built with pale conjunctivae. Oral examination revealed pooling of secretions, glossitis and angular cheilosis. The posterior wall of pharynx was seen normal and the gag reflex was intact. Examination of his extremities showed pale fingernail beds with koilonychias and absent palmar flexion creases. The mental functions of the patient were within normal limits and he has no other physical abnormalities. Laboratory report showed Hb%–7gm/dl, serum iron–20 \( \mu \)g/dl and red cell count–3.38 millions/\( \mu \)l. Laboratory report was in favor of profound iron deficiency anemia. Upper gastrointestinal (GI) endoscopy was done, showing an obstructing esophageal web (Fig. 1). The remainder of the upper endoscopic examination was unremarkable. Barium swallow showed a web in the upper part of esophagus (Fig. 2). The patient’s presentation of dysphagia, iron deficiency anemia and esophageal web in upper GI endoscopy fulfills the triad for PVS. Giemsa Trypsin/Leishman (GTL) banding technique was used for cytogenetic assessment as a predictor of risk of development of PCC in PVS. The assessment was carried out by detecting any chromosomal aberrations, but our case does not reveal any abnormal chromosomes (Fig. 3), which is responsible for PCC. The man was treated with endoscopic dilatation and iron supplementation and now he is in good health since one year after the treatment.

3. Discussion

Plummer–Vinson syndrome was first described in early part of 20th century as a triad of dysphagia, esophageal web and iron deficiency anemia. Plummer [5] established the syndrome for the first time in 1912 and has reported diffuse dilation of the esophagus and spasm of the upper esophagus without anatomical stenosis. Paterson [6] and Kelly [7], Otolaryngologists of Scotland reported a link between the anemia and dysphagia. Subsequently, Vinson [8] who was Plummer’s pupil, also reported a relation between the dysphagia, the anemia with angulation of the esophagus.

The exact etiology of the PVS remains controversial, but it has been associated with nutritional deficiencies, autoimmune disorders, and hereditary factors with a remarkably high female to male ratio [9]. It is reported that iron deficiency leads to a decrease in iron-dependent oxidative enzymes, which results in gradual degradation of muscles of the pharynx. As
a result, mucosal atrophy leads to web formation [10] and development of neoplastic changes at the lower part of the pharynx and upper esophagus [11]. It is seen that esophageal motility is also decreased in PVS. New motility studies showed normal amplitude of contraction after iron therapy [12]. Genetic transmission has gained little acceptance to date. But some families and their relatives have been seen as having this syndrome. Furthermore, PVS and sideropenic dysphagia are rarely seen among blacks [10].

Patients with PVS are diagnosed on the basis of thorough history taking, general and Ear, Nose and Throat (ENT) examination, laboratory investigations like hemoglobin (Hb) level, serum iron, iron binding capacity, plain lateral X-ray film on the neck and barium swallow in those with progressive dysphagia, direct endoscopic examination and biopsy taking for histopathological examination. The esophageal webs can be detected radiologically but small ones are usually overlooked, unless directly seen via endoscope [13].

PVS can be treated with iron supplementation and mechanical dilation. In case of severe esophageal web with significant narrowing and persistent dysphagia, patient needs rupture and dilatation of the web along with iron supplementation.

As PVS is associated with high risk of squamous cell carcinoma of the pharynx and the esophagus, the patients should always be followed closely. Cytogenetic assessment is a very good predictor of the possibility of PCC in patients with PVS. The chromosomal analysis revealed aberrations in 1q21, 10q11.2-1, 12p13, 15q22, 19q13 and Xq28 which are responsible for the PCC [3]. The chromosomal aberrations occur in the form of losses, gains, duplication, breakpoints and translocations. Our case did not reveal any chromosomal aberrations which rule out the development of any malignant changes. Esophageal webs in PVS are concerning not only symptomatically, but also are associated with increased risk of alimentary tract carcinoma [14]. For this reason, screening with upper GI endoscopy and chromosomal analysis are advised to detect early transformation.

4. Conclusion

Plummer–Vinson syndrome (PVS) or Paterson–Kelly syndrome presents as classical triad of dysphagia, esophageal web and iron deficiency anemia. Exact epidemiological data of this syndrome are not available; the syndrome is rarely encountered now a days. PVS reports have become rare in recent years, with occasional cases reported infrequently. The incidence of the PVS is extremely rare among Indian males in particular. Cytogenetic assessment is a very good predictor to rule out early detection of PCC in patients with PVS. So screening with upper GI endoscopy and chromosomal analysis is advised to detect early malignant transformation in PVS.

Conflict of interest

The authors declare no conflict of interests.

References


