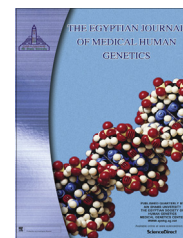




Ain Shams University
The Egyptian Journal of Medical Human Genetics

www.ejmhg.eg.net
www.sciencedirect.com



CASE REPORT

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



Santosh K. Swain ^{a,*}, Rajalaxmi Panigrahy ^b, Mahesh C. Sahu ^c

^a Department of ENT, IMS and SUM Hospital, Siksha “O” Anusandhan University, K8, Kalinganagar, Bhubaneswar 751003, Odisha, India

^b Department of ENT, Hi-Tech Medical College, Odisha, India

^c Central Research Laboratory, IMS and SUM Hospital, Siksha “O” Anusandhan University, K8, Kalinganagar, Bhubaneswar 751003, Odisha, India

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.

© 2015 The Authors. Production and hosting by Elsevier B.V. on behalf of Ain Shams University. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

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, web formations and malignant changes [4]. PVS is also associated with koilonychia, phagophagia and dysphagia due to 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

* Corresponding author. Mobile: +91 9556524887.
E-mail address: swainsantoshbbsr@yahoo.com (S.K. Swain).
Peer review under responsibility of Ain Shams University.

post-cricoid carcinoma (PCC), chromosomal assessment was done after taking patient consent.

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 µg/dl and red cell count–3.38 millions/µl. 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

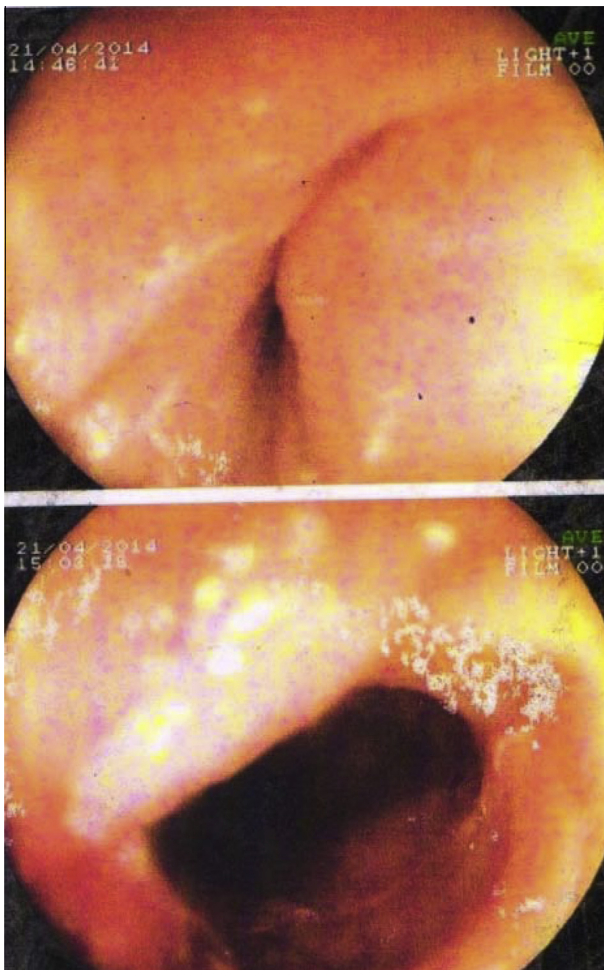


Figure 1 Upper gastrointestinal (GI) endoscopy showing an obstructive web at upper esophagus.

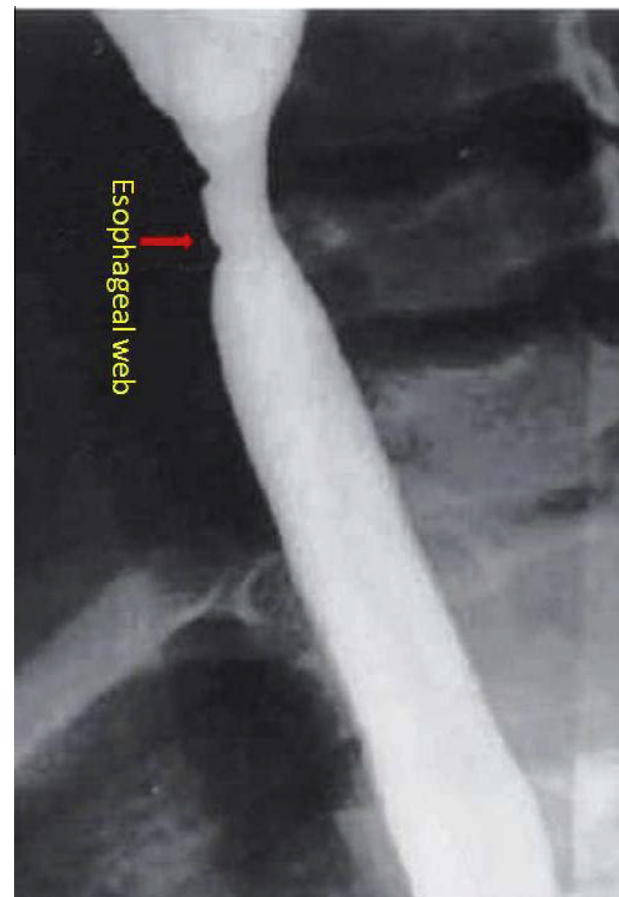


Figure 2 Barium swallow showing a web at upper esophagus.

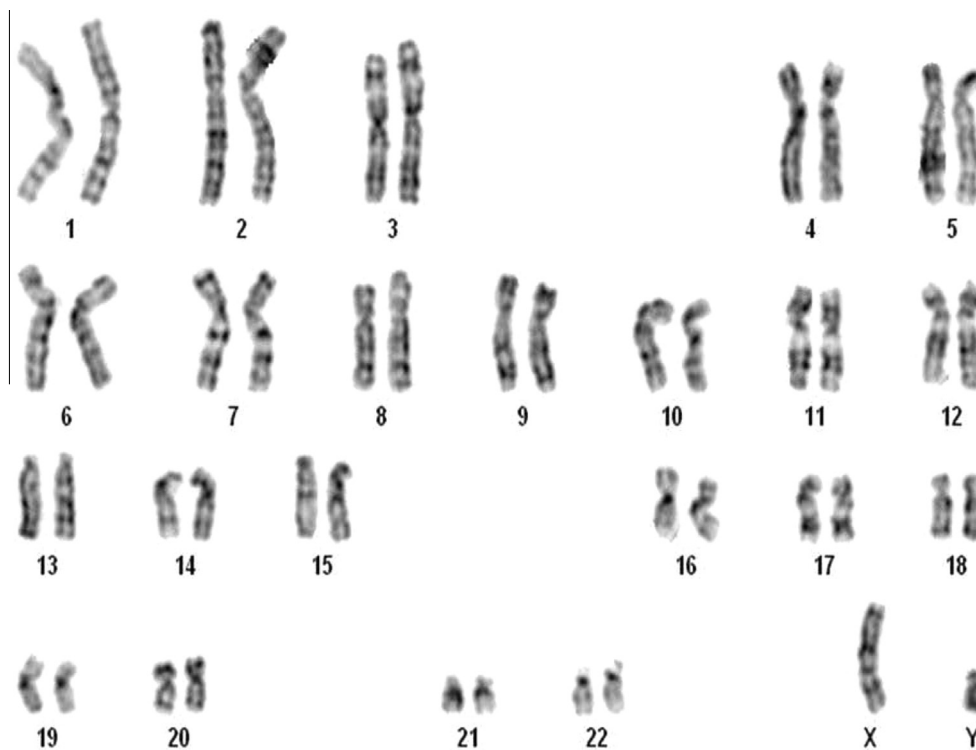


Figure 3 Chromosomal analysis of the patient.

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 $1q^{21}$, $10q^{11-11.1}$, $12p^{13}$, $15q^{22}$, $19q^{13}$ and Xq^{28} 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

- [1] Sinha V, Prajapati B, George A, Gupta D. A case study of Plummer–Vinson Syndrome. *India J Otolaryngol Head Neck Surg* 2006;58(4):391–2.
- [2] Novacek G. Plummer–Vinson syndrome. *Orphanet J Rare Dis* 2006;1:36.
- [3] Habib TN, Badawy BS. Cytogenetic assessment as a predictor of risk for poststeroid carcinoma associated with Plummer–Vinson syndrome. *EJNSO* 2015;2(1):1–5.

- [4] Donhue-Cornejo A, Guzman-Gastelum DA, Constandse-Cortez D, Gaitan-Cepeda LA, Escalera CR. Squamous cell carcinoma in the tongue and Plummer–Vinson syndrome – a case report. *Rev Odontol Mex* 2011;15:189–92.
- [5] Plummer S. Diffuse dilatation of the esophagus without anatomic stenosis (cardiospasm): a report of 91 cases. *J Am Med Assoc* 1912;58:2013–5.
- [6] Paterson DR. Clinical type of dysphagia. *J Laryngol Otol* 1919;34:289–91.
- [7] Kelly AB. Spasm at entrance of esophagus. *J Laryngol Otol* 1919;34:285–8.
- [8] Vinson PP. A case of cardiospasm with dilatation and angulation of the esophagus. *Med Clin Am* 1919;3:623–7.
- [9] Wynder EL, Hultberg S, Jacobsson F, et al. Environmental factors in cancer of the upper alimentary tract: a Swedish study with special reference to Plummer–Vinson (Paterson–Kelly) syndrome. *Cancer* 1957;10:470–82.
- [10] Atmadzitis K, Papaziogas B, Pavlidis T, et al. Plummer–Vinson syndrome. *Dis Esophagus* 2003;16(2):154–7.
- [11] Anderson SR, Sinacori JT. Plummer–Vinson syndrome heralded by postcricoid carcinoma. *Am J Otolaryngol* 2007;28:22–4.
- [12] Demirci F, Savas MC, et al. Plummer–Vinson syndrome and dilation therapy: a report of two cases. *Turk J Gastroenterol* 2005;16(4):224–7.
- [13] Oranu A, Buchholz M, Okafor N, Scott J, Henderson E. Unusual case of dysphagia in a male. *J Natl Med Assoc* 2010;102(9):832–4.
- [14] Jessner W, Vogelsang H, Puspok A, et al. Plummer–Vinson syndrome associated with celiac disease and complicated by postcricoid carcinoma and carcinoma of the tongue. *Am J Gastroenterol* 2003;98:1208–9.