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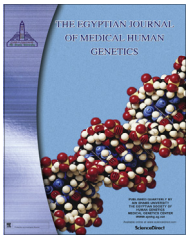

CASE REPORT

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

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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.

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

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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.

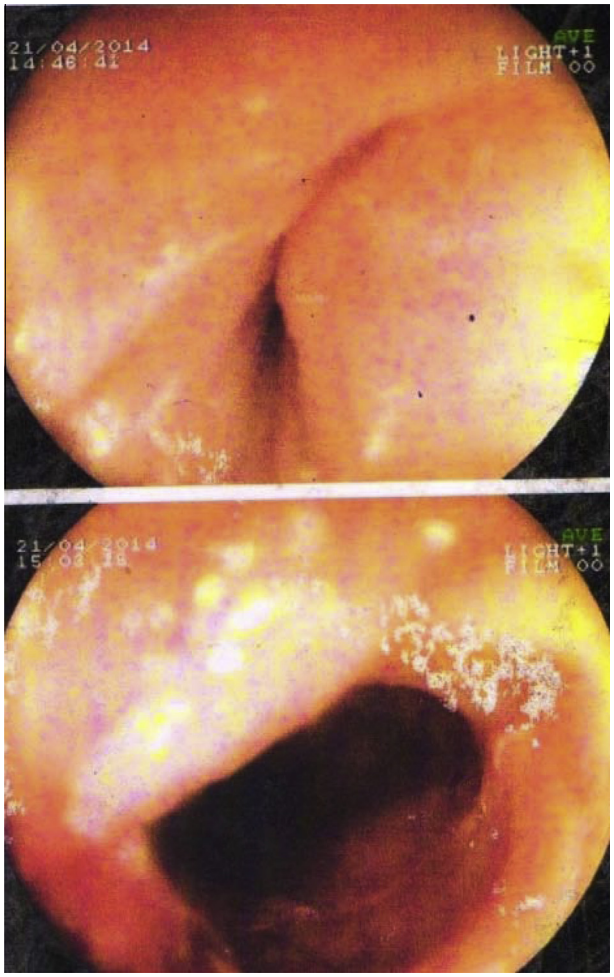

Figure 1 Upper gastrointestinal (GI) endoscopy showing an obstructive web at upper esophagus.

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

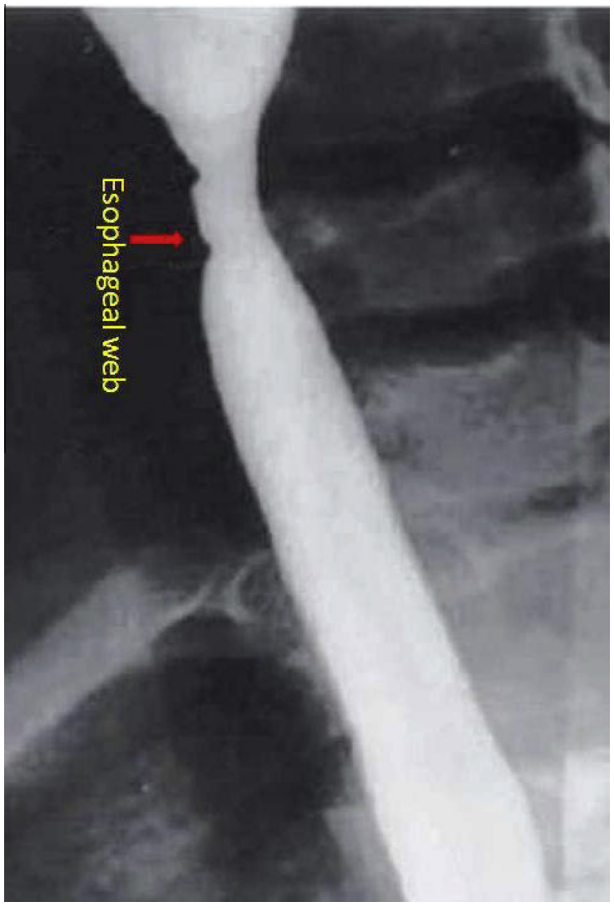

Figure 2 Barium swallow showing a web at upper esophagus.