Case Report

Extragonadal yolk sac tumor following congenital buccal mature cystic teratoma

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Abstract

Yolk sac tumor (YST) and teratoma both categorized as germ cell tumor (GCT). YST shows preferential differentiation toward yolk sac structures, while teratoma consists of tissues that originate from at least two embryonic germinal layers. Extragonadal location of YST is rare, whereas extragonadal teratoma is majority presented in nasopharynx area. Mature teratoma tends to be benign although some malignant transformation can occur. Recurrence of teratoma was reported mostly in the case of immature teratoma. YST occurrence after removal of mature teratoma is never reported. It is extremely rare for a second GCT to occur at the same site and with a different histological type. We herein report a case of a female infant presented with YST following a congenital buccal mature teratoma.

Keywords: Buccal teratoma, extragonadal yolk sac tumor, mature teratoma

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INTRODUCTION

Yolk sac tumor (YST) is defined as tumor that shows preferential differentiation toward yolk sac.[1] It is one of the most common tumors in pediatrics, usually arises in gonads. Extragonadal location is rare.[2,3] YST developed after teratoma is usually associated with immature teratoma.[2,4] It is extremely rare for a second germ cell tumor (GCT) to occur with a different histological type after removal of mature teratoma.[1,3] We report a case of female infant presented with YST following removal of a congenital buccal mature cystic teratoma. To our knowledge, this is the first case to be published in English literature.

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CASE REPORT

A 2-day-old infant girl was referred to emergency room for dyspnea due to a congenital oral mass obstructing her airway. Physical examination showed the mass-occupying oral cavity, protruding from left buccal mucosa, passing the midline of palates. The mass was similar in color as its surrounding tissue with smooth surface and cystic consistency. It was not attached to palates or base of tongue. Tracheostomy was performed immediately to secure her airway. Laboratory examination and chest X-ray were in normal range. Angiography showed neither stenosis nor malformation of blood vessels in carotid, cerebral and vestibulobasilar arteries. At the age

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of 3 weeks, she underwent surgery to remove the mass. Gross feature of the resected mass was an oval-shaped gray-brown tumor measured 4 cm × 2 cm × 1 cm. On the maximum cut surface, the tumor consisted of cystic space. The specimen was carefully sliced and submitted in its entirety to be examined. Microscopically, the tumor was composed of benign looking tissues including skin, glial tissue, cartilage and intestine. No immature component was observed. Histopatological diagnosis was mature cystic teratoma [Figure 1]. The infant was born at term, normally, weighing 3100 g and 49 cm, with no records of abnormal prenatal ultrasound examination.

Fifteen months after surgery, the patient started to have difficulty in feeding. Physical examination showed that a new mass had developed at her buccal mucosa, same site of her previous tumor and palate. This mass was tender on consistency. Computed tomography (CT) scan image showed that the tumor consisted of predominantly lipid component, such that clinical diagnosis was made to be lipoma, with lipoblastoma and teratoma as differential diagnosis. She then underwent the second surgery. The resected specimen was a white and brown tumor measuring 3 cm × 3 cm × 1 cm and firm in consistency. Approximately 4 cc of shredded tissue was also sent. All tissue received were embedded and processed for histological examination. Microscopically, the tumor consisted of tissue in polypoid shape, lined by stratified squamous epithelial cells with some edematous stroma. There was also part of tumor that consisted of mature fatty cells. No sign of malignancy was found [Figure 2].

One month after second surgery, a mass developed around the site of previous surgery scar. This mass grew rapidly, reaching 8 cm in diameter at 1 month. On physical examination, the mass was 8 cm × 6 cm × 5 cm in size, cystic with some solid area in consistency. CT imaging showed low-density tumor involving base of skull and mastoid bone. Chest X-ray was normal. CT abdomen and whole-body scan revealed no metastasis

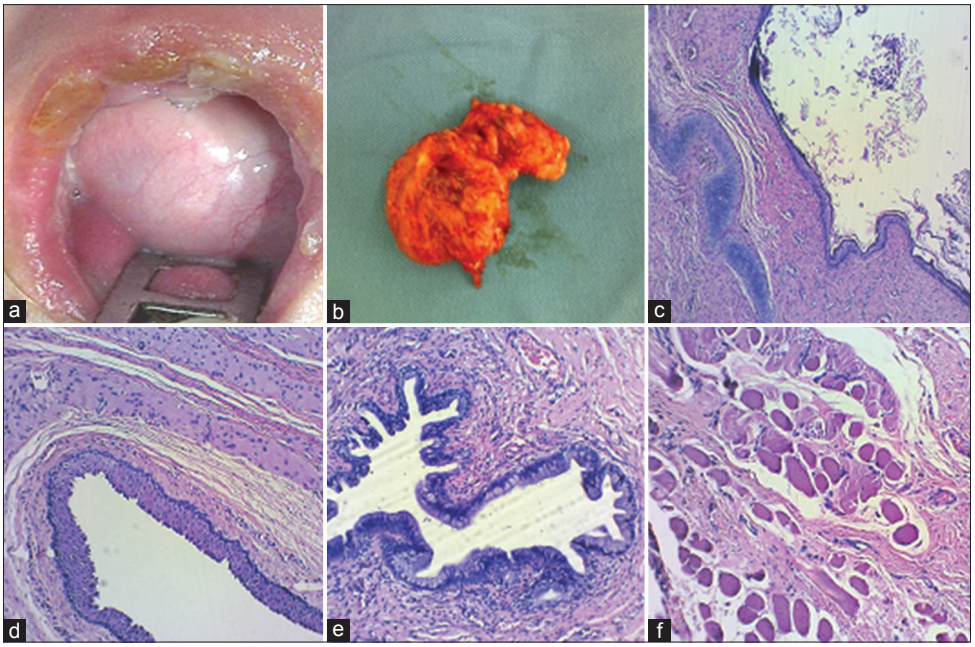

Figure 1: First presentation. (a) Congenital buccal mass on examination, (b) gross appearance of resected mass. Microscopically mass showing (c) cartilage and epidermal cyst, (d) glial tissue and urothelium, (e) gastrointestinal tract epithelium, (f) striated muscles. (b-f, H and E, ×4)

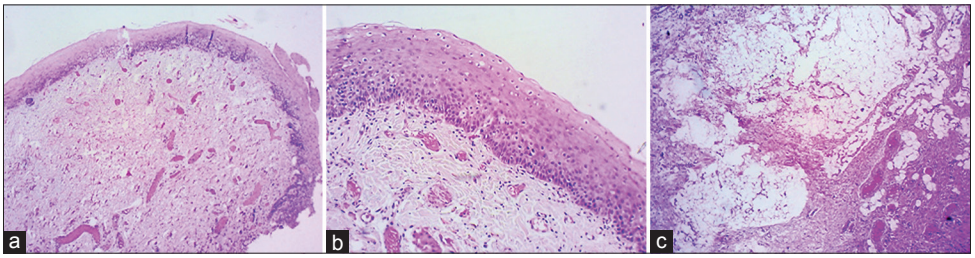

Figure 2: Second presentation. Resected mass from second surgery revealed structures microscopically (a) polypoid shaped mass line lined by epithelium (H and E, ×4), (b) squamous epithelium lining the polyp (H and E, ×10), (c) stroma with mature fatty cells (H and E, ×4)

foci. A third surgery was performed, resulting a tumor mass measuring 7 cm × 4 cm × 2.5 cm, brown in color and firm in consistency. Microscopically, the tumor showed proliferation of immature cell having clear nuclei, forming trabecular-papillary-alveolar structure. Some Schiller-Duval bodies, hyaline globule, area of necrosis and hemorrhage were also seen. Histopathological diagnosis made was YST [Figure 3].

Immunohistochemical staining from paraffin section showed strong positivity for alpha-fetoprotein (AFP), pan-cytokeratin (AE1/AE3) and anti-glypican 3 [Figure 3]. The serum AFP level after surgery was 351 ng/mL. Re-evaluation of teratoma slide was done, and the same result came up.

DISCUSSION

YST and teratoma both categorized as GCT. The incidence of GCT throughout life increases at two peaks of onset: peak one on between birth and 4 years of age and peak two on the second decades of life.[5] The most GCT in neonatal period is teratoma. The ratio of sex was 3:1 for females to male predominance.[5] GCT can arise in gonads and extragonadal setting. In the newborn age of group, the majority of cases are extragonadal.

Extragonadal of YST is rare compared to teratoma. It mainly presented in the central nervous system. The presence of YST in head and neck region is even more rare, but previously has been reported in orbit, postauricular region, parapharyngeal space, nasal cavity and floor of the mouth.[6,7,8] The rarity of its occurrence in head and neck area made YST at on the last possibility of diagnosis, thus making diagnosis difficult, especially if histological appearance was not classic.

YST developed after teratoma is usually associated with immature teratoma;[2] however previous and this present case proved that it can also develop following a mature teratoma.[3,9]

Congenital teratoma, especially one with large size, can be detected in prenatal checkup using ultrasound examination. Severe respiratory distress due to airway obstruction or displacement by the tumor is the most fatal postnatal complication. Prenatal diagnosis is very critical, allowing early appreciation of the mass that obstructs the airways.[10]

Our case presented with congenital mature teratoma that was located in the buccal mucosa of oral cavity. The location of mass, age of the patient and her clinical presentation had raised the issue of life-threatening airway obstruction. Delay in proper and prompt management of airway obstruction can cause death, especially in infants.

The etiology and natural course of teratoma have not been fully understood, but there are three hypotheses that have been postulated to explain it. First is the hypothesis that suggests that teratoma arises from totipotential primordial germ cells. These germ cells are located in the embryo in the wall of the yolk sac and migrate to the gonadal ridges during week 4 or 5 of gestation. The aberrant migration may give rise to teratoma, especially in extragonadal setting.[10] The second hypothesis suggests that teratoma originates from the remnants of the primitive node. However, the last hypothesis sees teratoma as a form of incomplete twinning, the same concept with fetus *in fetu*.[11]

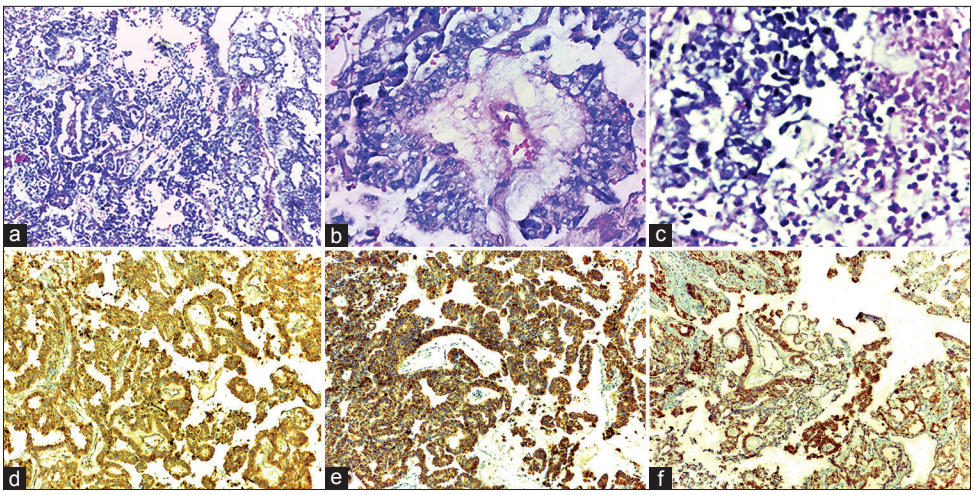

Figure 3: Third presentation. Microscopic appearance showing (a) trabecular-papillary-alveolar pattern (H and E, ×4), (b) Schiller-Duval bodies (H and E, ×40), (c) hyaline globules (H and E, ×20). Immunohistochemical staining showed strong positivity for (d) alpha-fetoprotein, (e) pancytokeratin (AE1/AE3) and (f) glypican 3

The exact mechanism of recurrence of mature teratoma has not been well established, especially in terms of why mature teratoma recurrence as YST as like in the present case. Some investigators proposed that YST could develop directly from teratoma via malignant transformation, whereas others suggested that YST would develop from teratoma that contains small foci of YST that were not recognize initially.[3,12,13] Yoshida *et al.* proposed that recurrence of YST after mature teratoma excision was caused by residual teratoma cells that still remained in the border or tumor area postsurgically, with or without YST foci.[14] Thus, this theory regards the recurrence as a form of the tumor relapse.

Surgical procedure remains the treatment of choice for teratoma, regardless of location. Compared to gonadal teratoma, extragonadal setting of disease may challenge the procedure of surgery. Extirpation of teratoma in the head and neck area requires more advanced technical skill and raise the issue of severe surgery complication due to its proximity to vital organs such as major blood vessel, especially in neonates. In general, the resection should be done completely with clear margin from cell tumors.[15] This margin issue is related to the possibility of recurrence of the tumor after resection although it is debatable.

Recurrence can occur shortly after primary teratoma, or in a long-term up until 7 years after initial tumor, and can appear in different histological types of tumor-like YST.[10,15] Careful long-term follow-up through clinical examination, serum level evaluation and MRI is needed.[13-15]

Certain type of GCT secretes proteins that can be used as markers of tumor presence. Elevated AFP serum level is a common finding in patients with YST. In this present case, AFP serum after surgery was 351 ng/mL. It was not considered extreme, but it showed marked elevation even though the tumor was already taken out.

There is some differential diagnosis of YST including juvenile granulosa cell tumor (JGCT) of sex-cord stroma group, dysgerminoma, and embryonal carcinoma (EC).[1,14] JGCT does not cause elevated serum AFP level. Microscopically, the solid and macrofollicular pattern of JGCT can resemble the solid, macrocytic and polyvesicular vitelline pattern of YST. Immunohistochemically, YST shows strong positivity for AFP, cytokeratins and glypican-3. However, JGCT usually shows positive staining for sex-cord stromal tumor markers such as inhibin and calretinin.[6,7,14,15] When YST appears in solid patterns, it can mimic dysgerminoma. Dysgerminoma cells are larger and have medium to large vesicular nuclei with clear chromatin and obvious nucleoli. Immunohistochemical staining can help differentiate these two. Dysgerminoma will stain positive for CD117 and D2-40 and negative for keratins, AFP and glypican-3.[15] EC and YST have overlapping appearance in morphologies that can be hard to be distinguished unless specific patterns of YST are observed. Immunohistochemical staining for EC will be positive for OCT4, Nanog, Sox-2 and CD 30 and negative for glypican-3. AE1/AE-3 will show membranous staining in EC and cytoplasmic staining in YST. Some EC cells can be positive for AFP, but strong positivity favors for YST.[6,14,15]

Histologically, this present YST case showed immature cells with clear nuclei, forming trabecular-papillary-alveolar structures. The appearance of Schiller-Duval bodies, that are considered pathognomonic for YST, assures the diagnosis.[6] Immunohistochemical staining of our case showed strong positivity for AFP, pan-cytokeratin (AE1/AE3) and anti-glypican 3, further confirming the diagnosis of YST.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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