Trichoepithelioma is a rare benign skin lesion that originates from hair follicles. Trichoepitheliomas are mostly cylindromatosis, and multiple familial trichoepithelioma. Result in one of 3 syndromes: Brooke-Spiegler syndrome, familial cases appear to be related to a mutation in a gene encoding a tumor suppressor located on band 9q21. Additionally, the gene involved in basal cell carcinoma (PTCH, human patched gene located on band 9q22.3) appears to participate in the pathogenesis of trichoepithelioma. Brooke-Spiegler syndrome patients have a high incidence of multiple skin appendage tumors such as cylindroma, trichoepithelioma, and spiradenoma. These patients may show mutations of the CYLD gene (cylindromatosis gene) that map to 16q12-13.

A 2006 study has suggested that abnormalities in this gene may result in one of 3 syndromes: Brooke-Spiegler syndrome, familial cylindromatosis, and multiple familial trichoepithelioma. A 2009 study reports a novel missense mutation in the CYLD gene, heterozygous nucleotide G→A transition at position 2,317 in exon 17, in a Chinese family with multiple familial trichoepithelioma. Additionally, a novel splicing mutation in the CYLD gene (IVS12 + 1 G→A) has been reported in a Taiwanese family with multiple familial trichoepithelioma. The gene associated with the familial type of trichoepithelioma links to the short arm of chromosome 9. Because several tumor suppressor genes (ie, p16, p15, and the gene for the basal cell nevus syndrome) are in this region, the gene for the development of familial trichoepithelioma also encodes for a tumor suppressor. If altered, cellular proliferation may be up-regulated because of a poorly functioning or absent tumor suppressor. Studies have indicated that CYLD encodes a deubiquitinating enzyme that negatively regulates the nuclear factor (NF)-κappaB and c-Jun N-terminal kinase (JNK) pathways. Due to the presence of significant numbers of Merkel cells within the tumor nest and the detection of a sheath of CD34-positive dendrocytes around the tumor nests, it appears that trichoepithelioma differentiates toward or derives from hair structures, particularly the hair bulge. Rare instances of tumors resembling trichoepithelioma have been reported in animals.

Familial cases appear to be related to a mutation in a gene encoding a tumor suppressor located on band 9q21. Additionally, the gene involved in basal cell carcinoma (PTCH, human patched gene located on band 9q22.3) appears to participate in the pathogenesis of trichoepithelioma. Brooke-Spiegler syndrome patients have a high incidence of multiple skin appendage tumors such as cylindroma, trichoepithelioma, and spiradenoma. These patients may show mutations of the CYLD gene (cylindromatosis gene) that map to 16q12-13.

Case Details
A twenty-four(24) years old Indian lady presented to our hospital with history of scalp swelling for two years. The swelling was insidious in onset with initial size of about 2 x 2 cm. It was gradually increasing in size. There was rapid increase in size in the last 6 months. History of ulceration over swelling present from 2 weeks. There were no other swellings elsewhere in the body.

On physical examination, she was moderately built and nourished. Vital signs were normal. Local examination revealed a 9 x 8 cm swelling in right parietal region, irregular in shape, with ulcerations over the skin. Surrounding skin was normal. There were no signs of inflammation. It was firm in consistency and did not appear to be fixed to underlying skull. No regional lymph nodes palpable. Other systemic examination was unremarkable.

Laboratory tests were unremarkable. Plain skull X-ray did not reveal involvement of skull. Biopsy of the lesion was suggestive of trichoepithelioma with suspicion of malignancy.

On surgical intervention the growth was found to infiltrate the periosteum. Wide local excision of the tumor was done; along with excision of outer table of skull.

Defect was covered using locally advanced flap and skin grafting.
Discussion
Skin adnexal tumours (SAT) are a large and diverse group of benign and malignant neoplasms, which exhibit morphological differentiation towards one of the different types of adnexal epithelium present in normal skin: pilosebaceous unit, eccrine and apocrine. SAT may display more than one line of differentiation (hybrid/composite tumours), rendering precise classification of these neoplasms difficult. The diagnosis of these mixed SAT relies on histological evaluation, and they are usually classified according to the predominant morphological component. The histogenesis of mixed adnexal tumours is still uncertain; however, the possibility of origin from a pluripotent stem cells is suggestive.

Most SAT are benign, and local complete surgical excision is curative. However, diagnosing some of these tumours has important implications, as they might be markers for syndromes associated with internal malignancies, such as trichilemmomas in Cowden disease and sebaceous tumours in Muir–Torre syndrome. A malignant counterpart of almost every SAT has been described. These tumours are rare, locally aggressive, and have the potential for nodal involvement and distant metastasis, with a poor clinical outcome. Therefore, establishing a diagnosis of malignancy in SAT is important for therapeutic and prognostic purposes. A vast variety of differential diagnosis has to be kept in consideration while treating such scalp tumours viz

- Basal cell carcinoma
- Microcystic adnexal carcinoma
- Trichoadenoma
- Tumor of follicular infundibulum
- Basaloid follicular hamartoma

To conclude, a very high index of suspicion and a competent pathological support is mandatory and highly beneficial in the diagnosis of this rare scalp tumor entity.

FIGURES

FIGURE 1

FIGURE 2

FIGURE 3

FIGURE 4

FIGURE 5

References