Retinal astrocytic hamartoma and systemic spectrum of disease in successive generations with tuberous sclerosis complex

Gitanjli Sood, Ramanuj Samanta, Sneha Pandurangan, Pankaj Sharma, Anupam Singh, Ajai Agrawal

Key words: Adenoma sebaceum, ash leaf macule, cortical tuber, retinal astrocytic hamartoma, tuberous sclerosis

A 26-year-old female presented with recent burning micturition and few skin lesions since teenage. Dermatological evaluation revealed adenoma sebaceum [Fig. 1a] and ash leaf spots [Fig. 1b and c] over trunk and abdomen. Her best-corrected visual acuity was 20/20 in both eyes. Dilated fundus showed multiple flat translucent whitish-patches along the supero-temporal arcade of right eye suggestive of retinal astrocytic hamartoma (RAH; Fig. 1d). Corresponding Optical coherence tomography over these lesions showed hyperreflectivity of inner retinal layer [Fig. 1e]. Magnetic resonance imaging (MRI) of brain [Fig. 1f-i] revealed multiple cortical tubers in bilateral frontoparietal lobes. Contrast-enhanced computed tomography scan (CECT) revealed multiple renal angiomyolipoma [Fig. 1j] and cysts in kidney, ovary and liver [Fig. 1k]. Round to oval sclerotic lesions were also noted in spine, sacrum, and iliac bones [Fig. 1l].

Retrospective history revealed similar dermatological lesions in father and daughter of the affected female. Although her father was unavailable for investigation, 4-year-old daughter was evaluated for subnormal intelligence. Examination of daughter also revealed adenoma sebaceum and ash leaf spots [Fig. 2a and b], bilateral RAH [Fig. 2c and d], multiple cortical tubers [Fig. 2e-h], subependymal nodules [Fig. 2i-l] and renal abnormalities [Fig. 2m and n]. A definitive diagnosis of tuberous sclerosis complex (TSC) was made in both mother and daughter.[1] They were advised 6 monthly ocular follow-up and referred to general physician for systemic manifestations.

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Department of Ophthalmology, All India Institute of Medical Sciences (AIIMS), Rishikesh, Uttarakhand, India

Correspondence to: Dr. Ramanuj Samanta, Department of Ophthalmology, All India Institute of Medical Sciences (AIIMS), Rishikesh - 249 203, Uttarakhand, India. E-mail: ramanuj.samanta@gmail.com

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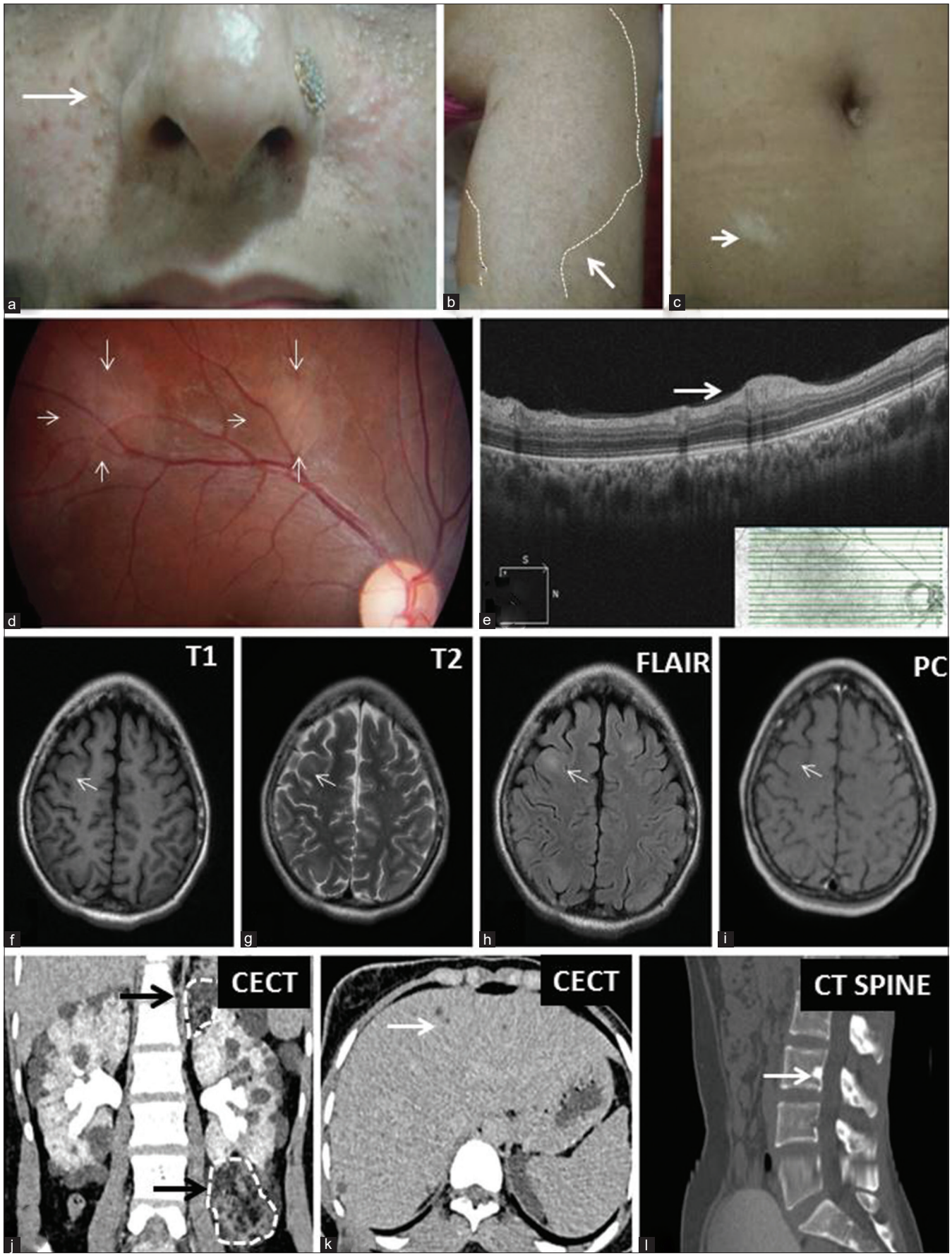

Figure 1: Clinical photograph (a-c) showing adenoma sebaceum (a) and ash-leaf macules (b and c). Fundus picture (d) showing multiple retinal astrocytic hamartomas (RAH) in right eye; OCT (e) showing hyperreflectivity of inner retinal layers. MRI brain (f-i) revealed cortical tubers in bilateral frontoparietal lobes. CECT abdomen (j and k) revealed enlarged kidneys, renal angiomyolipoma (j), and multiple cysts in kidney, ovary, and liver (k). Sclerotic bone lesion in spine (l) were also noted

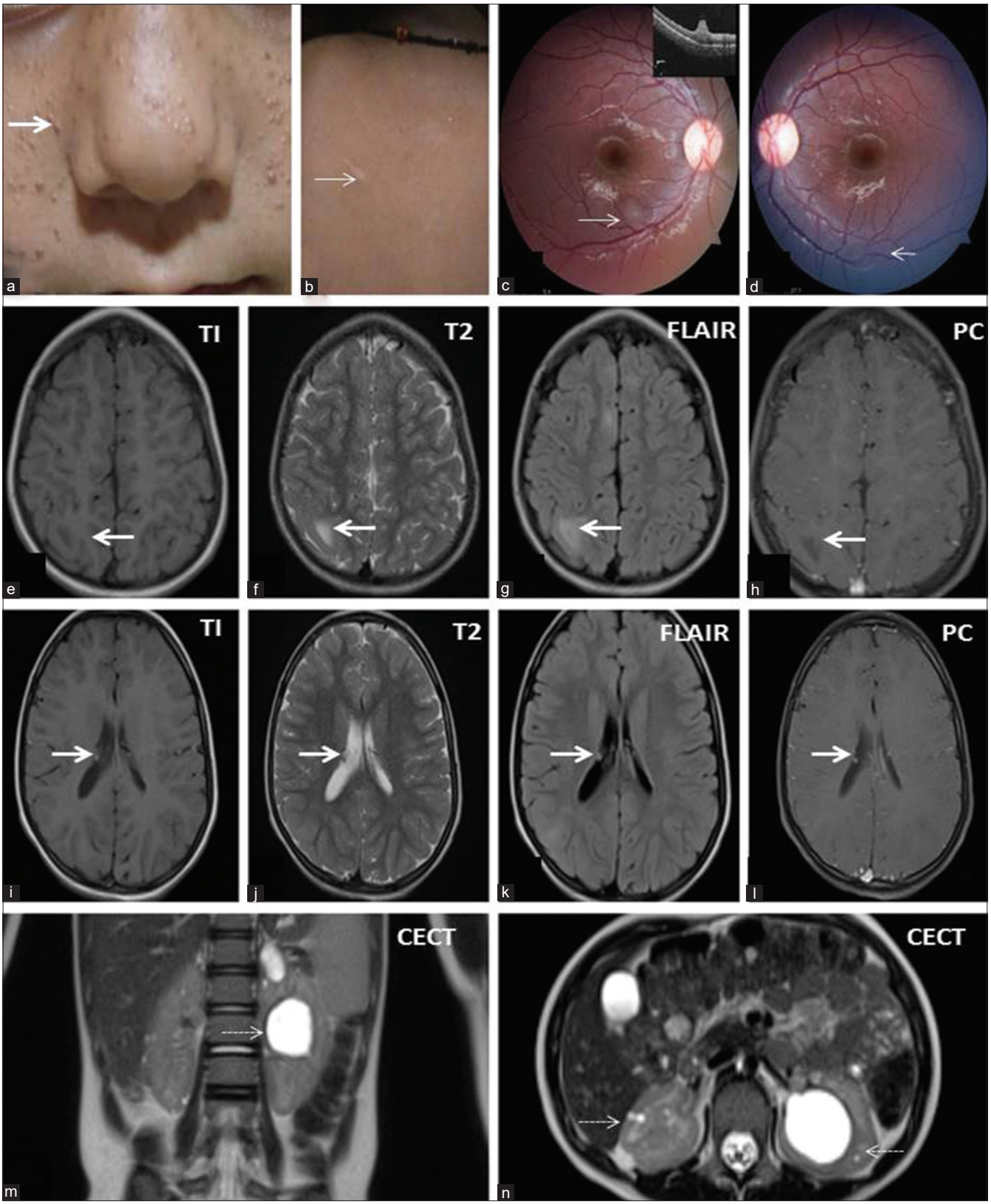

Figure 2: Clinical photograph (a and b) showing adenoma sebaceum (a) and ash-leaf macules (b). Fundus picture (c and d) of both eyes showing retinal astrocytic hamartomas (RAH). MRI brain revealed cortical tubers (e-h) and subependymal nodules (i-l). CECT showing dilated pelvicalyceal system (m) and multiple subcentimetric cysts in bilateral renal parenchyma (n)

Discussion

TSC is a rare phakomatosis characterized by multiple systemic hamartomas. It is inherited as autosomal dominant disorder with high penetrance and variable phenotypic expression.[2] RAH are primary ophthalmic features of TSC.[3] RAH are noted as subtle greying of retina and are often missed unless carefully searched. It can manifest morphologically as (a) translucent flat tumors, (b) whitish nodular (mulberry lesion) mass or (c) Mixed variety.[4] Dilated fundus evaluation is a simple and cost-effective way to clinically diagnose such disorders before other invasive investigations are undertaken to confirm TSC.

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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Nil.

Conflicts of interest

There are no conflicts of interest.

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