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A RARE PRESENTATION OF BRAIN CYSTIC DISEASE LINKED WITH ECTODERMAL DYSPLASIA (CEREBRUM POLYCYSTICA VERA): A NOVEL NEUROCUTANEOUS SYNDROME

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ABSTRACT

Neurocutaneous syndromes comprise a diverse group of conditions affecting both the nervous system and the integumentary system. Within this spectrum, we present a rare and compelling case of cerebrum polycystica vera associated with ectodermal dysplasia in a 30-year-old male. The patient's clinical journey includes a sequence of seizures progressing to left-sided weakness, accompanied by compelling dermatological manifestations. Extensive neuroimaging studies, including MRI and CT scans, reveal the presence of multiple cysts in the brain, highlighting the distinct nature of this novel neurocutaneous syndrome. Based on the authors' knowledge, this represents the first documented case report from Pakistan, while a few cases have been documented in international publications.

KEY WORDS: Cerebrum Polycystica Vera, Ectodermal Dysplasia, Neurocutaneous Syndrome, Seizures, Neuroimaging, Nervous system, Integumentary system.

INTRODUCTION

Neurocutaneous syndromes represent a diverse group of disorders characterized by concurrent involvement of the nervous system and the skin.[1] Among these, "cerebrum polycystica vera," a rare and emerging neurocutaneous syndrome, has gained attention due to its unique association with ectodermal dysplasia. This syndrome presents a compelling clinical challenge, as it underscores the complex interplay between the central nervous system and the integumentary system, rooted in their common embryological origins.

Ectodermal dysplasia, a congenital disorder that affects hair, teeth, skin, and nails, has been historically described in isolation. However, recent cases have shed light on a previously unrecognized connection between ectodermal dysplasia and the development of polycystic brain lesions, known as "cerebrum polycystica vera." [2] This intriguing association prompts further exploration into the shared embryonic origins of these seemingly disparate systems and its implications for both diagnosis and management.

CASE PRESENTATION

A 30-year-old male patient presented with a history of focal seizures originating from the left side, involving the left side of face and the body with subsequent generalization. These seizures occurred in a series of ten episodes over the span of a month, later he developed progressive left hemiparesis involving upper and lower limb simultaneously, rendering him bedridden in a week. It was not associated with numbness, dysphagia, dysarthria, facial deviation or urinary and fecal retention/incontinence. Notably, the patient's medical history revealed a unique childhood experience marked by recurrent skin rashes and nodular swellings with yellowish and black discharge. These cutaneous issues, affecting the head, face, neck, chest, abdomen, and back, had ultimately healed at that time with scarring. During that period, the patient's primary teeth were lost, and secondary teeth did not emerge. Severe plantar dryness and heel ulcers with bleeding, especially in winters were also documented. Past surgical history included two separate events of excision of a skin outgrowth on the lower back. No documented record was available. Family history was significant for notable palmar and plantar color changes in a sibling who had tragically faced sudden unexpected death in epilepsy, and also a twelve year-old niece, who recently had started developing similar changes.

On examination multiple plaque like irregular pigmented lesions with some nodules and residual healed scars accompanied by patchy areas of alopecia were seen on the head, neck chest, abdomen, back and feet. Figure 1 demonstrated the healed scars and alopecia.

Figure 1: Healed scars and alopecia in the patient

Oral cavity examination shows absent teeth except 2nd molar tooth in lower jaw. Dystrophic toenails and severe plantar and palmer dryness with ulceration were noted **(Figure 2).**

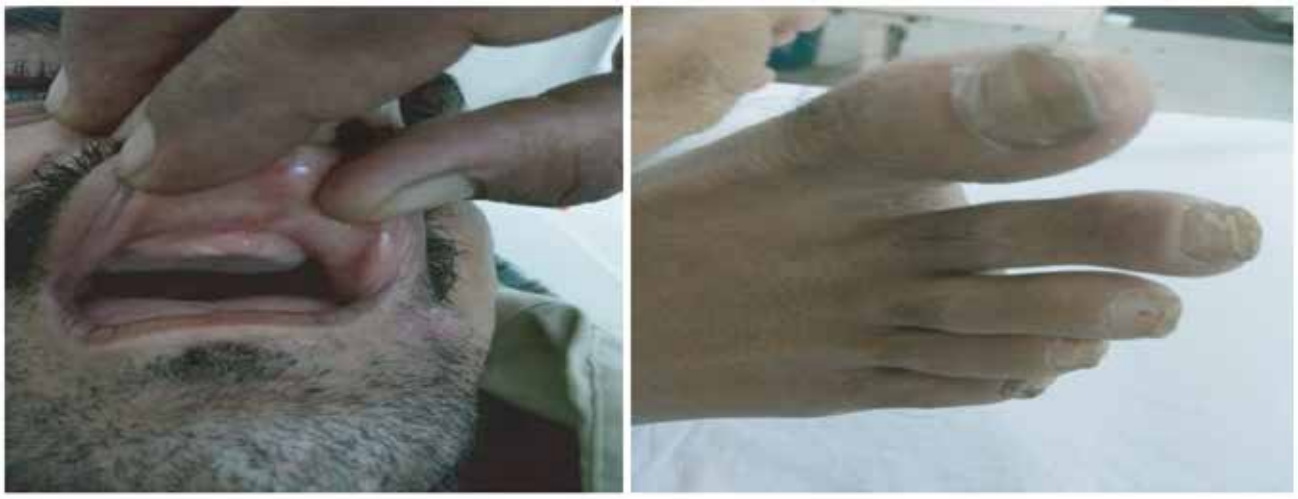

Figure 2: Oral cavity showing missing teeth and foot showing dystrophic toenails

On neurological examination, GCS was 15/15. Mini mental state examination (MMSE) showed mild to moderate cognitive decline. Fundoscopy showed bilateral chorioretinitis with old left chorioitic scar. On motor examination there was a noticeable reduction in muscle bulk in all four limbs. The patient exhibited a power of 0/5 in the left upper and lower limbs and 4/5 in the right upper and lower limbs. Reflexes were 3+ in the left and 2+ in the right, accompanied by an extensor plantar response on the left and a flexor plantar response on the right. Sensory, co-ordination and rest of examination was unremarkable.

A comprehensive diagnostic evaluation encompassed a range of investigations, including a complete blood count, biochemical analyses, liver function tests, renal function tests, viral markers, HIV screening, ANA and ENA profiling, Toxoplasmosis IgG, gastric lavage for AFB smear, culture, and gene expert analysis. All results returned normal/negative.

The patient's skeletal survey revealed a complex set of findings, including thickening and increased density in the cranial vault, absence of most teeth except for a single deciduous tooth, thinning of the mandible, paranasal sinus hypoplasia, osteopenia and diffuse cortical thickening in long bones and solid periostial reaction. The foot X-ray displayed a coarse trabecular pattern. These are shown in Figure 3.

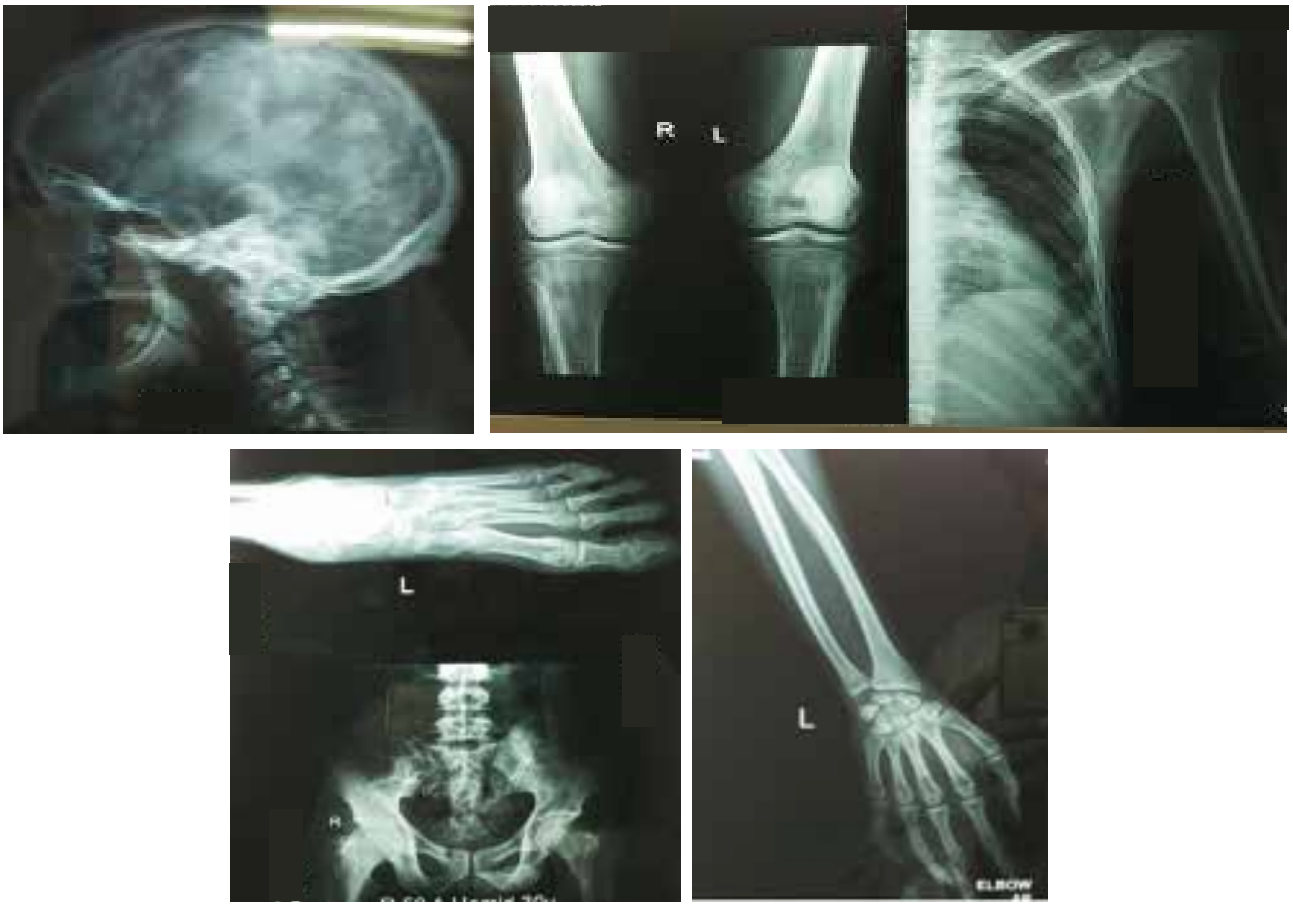

Figure 3: X-rays of the patient

CT scans of the brain showed multiple cysts with an area of extensive edema, causing mass effect in the right parietal region, compressing the lateral ventricle, and resulting in midline shift. Additionally, the cortical bone of the cranial vault displayed thickening with an irregular outline of the brain. The MRI further highlighted the extent of these cysts, including peripheral enhancement (Figure 4).

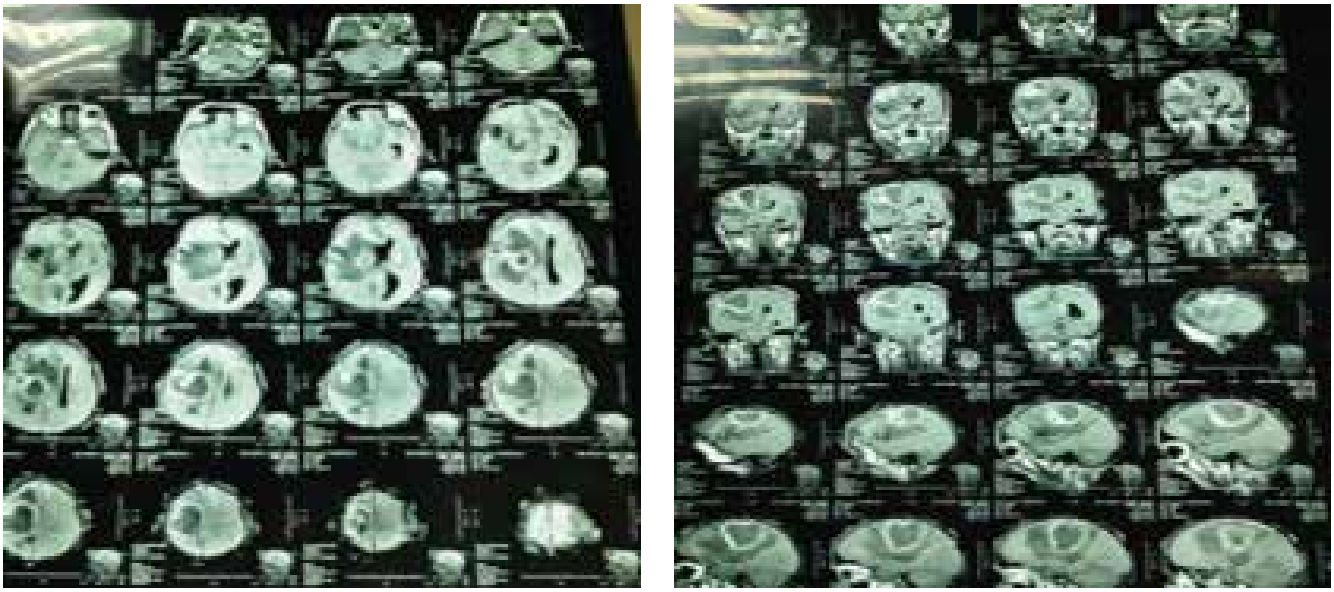

Figure 4: MRI brain showing contrast-enhancing cysts

Neurosurgical opinion was taken, they advised for open craniotomy after consultation with plastic surgeon about healing of wound and grafting. Attendants were counseled but they were not ready for craniotomy after explaining them about delayed wound healing and requirement for skin grafting.

The patient underwent a six-week inpatient stay. A treatment regimen included broad-spectrum antibiotics, offering coverage against viral, bacterial, and fungal pathogens. Additionally, steroids and antiepileptic medications were administered as part of the therapeutic protocol. The culmination of this treatment phase successfully controlled the patient's seizure activity, prompting a subsequent MRI evaluation after six weeks of therapy. On comparison to previous MRI, size of lesion was apparently the same but the edema had reduced grossly. Findings were suggestive of infected brain cysts in right parietal region (Figure 5).

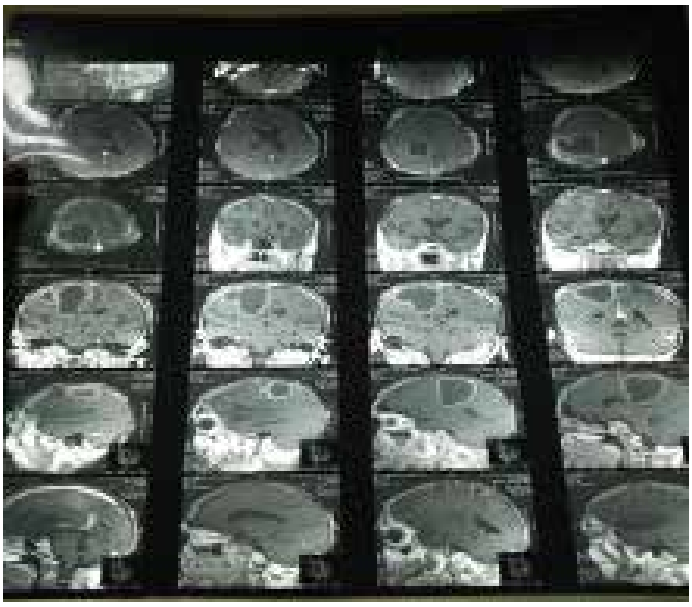

Figure 5: Follow up MRI showing reduction in the edema

DISCUSSION

It is postulated that a shared ectodermal origin may underlie the development of both the brain cysts and ectodermal dysplasia. This hypothesis is supported by the fact that various structures, including the central nervous system (comprising ependymal/leptomeningeal cells and retinal cells), the epidermis (encompassing hair and nails), and dental enamel, all originate from the embryonic ectoderm. This confluence of characteristics suggests the emergence of a novel and distinctive neurocutaneous syndrome.[3]

Ectodermal dysplasias can affect bones. These conditions primarily impact the skin, sweat glands, hair, teeth, and nails. When two or more body structures derived from the ectoderm are affected, a person is considered to have ectodermal dysplasia (ED). Specifically, the development of bones in the rest of the skeleton can be affected, leading to abnormalities in the long bones of the arms and legs (metaphysical dysplasia), resulting in short limbs and stature. Furthermore, the reported nervous system involvement in cerebrum polycystica vera typically involves dilated Virchow-Robin (VR) spaces in multiple lobes, whereas the patient in this report has a large right front parietal peripherally enhancing multiloculated cystic lesion. This raised the need of a biopsy but that was unfortunately refused by the family. However, this combination of neurological symptoms, family history, dermatological manifestations, and the presence of cystic brain lesions defines a complex and rare clinical presentation. The connection between ectodermal dysplasia and cerebrum polycystica Vera provides a unique perspective on the shared embryonic origins of these systems.[4]

CONCLUSION

This case underscores the unique convergence of neurological and dermatological symptoms, leading to the recognition of a novel neurocutaneous syndrome, cerebrum polycystica vera, associated with ectodermal dysplasia. This rare combination challenges established diagnostic and clinical paradigms, emphasizing the need for multidisciplinary assessments and genetic investigations to further our understanding of this condition.

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Sana Ghous; concept, case management, manuscript writing
Alam Ibrahim Siddiqui; case management, manuscript writing
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Anjlee Shankar; case management, manuscript revision
Asif Ali; case management, manuscript revision
Shaheen Bhatty; case management, manuscript revision
All the authors have approved the final version of the article and agree to be accountable for all aspects of the work.