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Case Report **OPEN ACCESS**

Cutis verticis gyrata and epilepsy, is there a typical patient?: A case report

Saleha Aldawsari [1], Mohammed Aljughayman [2], Almunthir Alhamed [3], Mohammed Alhazza [4], Nasser Almulhim [5*], Mohammed Alnaim [6], Farhan Siddiqui [7]

[1] King Fahad Hospital, Hofuf, SAUDI ARABIA
[2] Department of Dermatology, King Fahad Hospital, Hofuf, SAUDI ARABIA
[3] Department of Dermatology, Kind Saud University Medical City, Riyadh, SAUDI ARABIA
[4] Department of Neurology, King Fahad Hospital, Hofuf, SAUDI ARABIA
[5] Department of Dermatology, King Faisal University, Hofuf, SAUDI ARABIA
[6] Department of Neurology, King Faisal University, Hofuf, SAUDI ARABIA
[7] Department of Laboratory and Blood Bank, King Fahad Hospital, Hofuf, SAUDI ARABIA
***Corresponding Author:** almulhimnasser115@gmail.com

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ABSTRACT

Cutis verticis gyrata (CVG) is a rare skin condition characterized by thickened, folded scalp skin, which can occasionally coexist with neurological disorders such as epilepsy without typical known causes. It is important to be open to new explanations for this relationship. While certain characteristics are often seen in affected patients, the underlying reasons for this association remain unclear. Physicians should be aware that CVG could serve as an early indication of epilepsy or other neurological disorders. We report a case of a 15-year-old Saudi male with a history of epilepsy, who presented with progressive scalp swelling leading to the development of CVG.

Keywords: epilepsy, cutis verticis gyrata, neurology, dermatology

INTRODUCTION

Cutis verticis gyrata (CVG) is a rare benign cutaneous disorder characterized by thickening and folding of the scalp, resulting in a corrugated or ridged appearance akin to the brain's sulci and gyri. CVG is hardly ever encountered in practice as it only affects 1 in 100,000 men and 0.026 in 100,000 women. This condition has been categorized as either primary essential, primary non-essential or secondary. The secondary subtype has an underling pathological mechanism like genetic, endocrinological or inflammatory conditions while the primary subtypes have no such underling pathology. However, the primary non-essential CVG is differentiated from the primary essential CVG due to its association with neurological disorders. For example, CVG has been reported to be associated with epilepsy[1, 2].

Epilepsy is a chronic neurological disorder characterized by recurrent seizures. Seizures occur due to abnormal electrical activity in the brain, leading to temporary disruptions in behavior, consciousness, movements, or sensations [3]. There is no clear evidence to suggest a potential link between CVG and epilepsy, and the underlying mechanisms are not fully understood [4]. The coexistence of CVG and epilepsy can have significant impacts on the quality of life for affected individuals. The cosmetic appearance of CVG may cause psychosocial distress and affect self-esteem. Epilepsy, on the other hand, can result in limitations in daily activities, driving restrictions, and potential safety risks during seizures. Therefore, an integrated approach that addresses both the dermatological and neurological aspects of these conditions is crucial for optimal management and patient well-being.

CASE REPORT: MATERIALS AND METHODOLOGY

We present a case of 15 years old Saudi male, right-handed known case of epilepsy. He is the son of healthy non - consanguineous parents with no family history of psychomotor delay, epilepsy or CVG. There is an unremarkable antenatal history except for C section delivery and 3 days of pediatric intensive care unit admission due to respiratory distress. The patient had a normal and active childhood until he started to have difficulty speaking, change in personality and weakened academic performance at the age of 9 and was diagnosed with epilepsy. Initially he was given lacosamide with significant improvement, until it was unavailable, where he was shifted to levetiracetam and carbamazepine in 2021. During these years, the patient had episodes of visual hallucinations and dizziness which were resolved after the 3rd year of treatment. Afterwards, the patient showed great improvement, and the last documented seizure episode was 3 years before presentation. In the time of presentation, he was only prescribed levetiracetam once daily. One year before the patient was diagnosed with epilepsy, he complained of diffused swelling

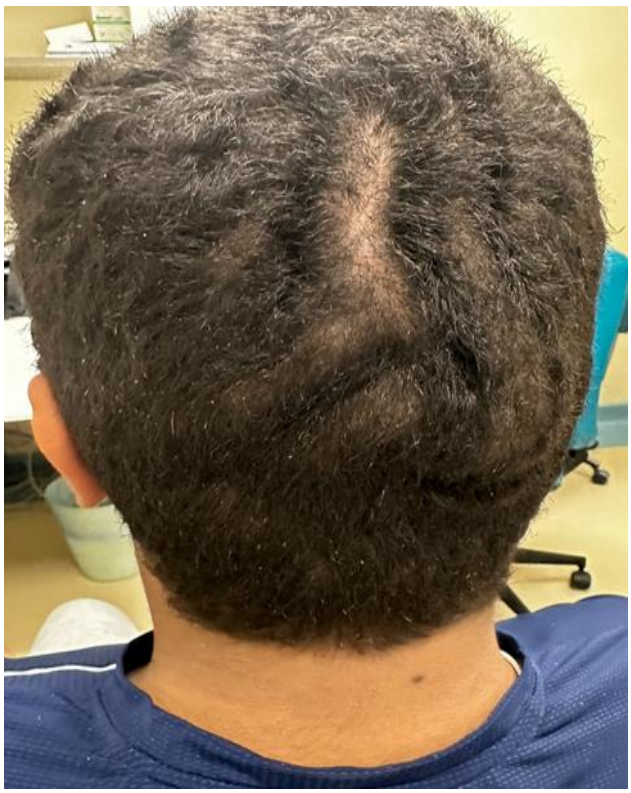

Figure 1. Multiple folds and furrows running in an anteroposterior direction on the parietal and occipital areas of the scalp (Reprinted with permission of patient)

Table 1. Results of laboratory investigations

Laboratory investigations	Result	Normal range
WBC	Normal	(4,000-10,000 cells/µL)
Hbg	High (18.2 g/dl)	(13.5-17.5 g/dL)
MCV	Low (75.7 fL)	(80-100 fL)
RDW-CV	High (16.3%)	(11.5-14.5%)
Lymphocyte	High (58.4%)	(20-40%)
Hepatitis serology	Negative	-
HIV	Negative	-
Prolactin	Normal	(4.0-15.2 ng/mL)
T3	Normal	(80-200 ng/dL)
T4	Normal	(4.5-12.5 µg/dL)
TSH	Normal	(0.4-4.0 mIU/L)
Growth hormone	Normal	(0-5 ng/mL)
Vitamin D	Low (15.8 mg/ml)	(30-100 ng/mL)
Testosterone	Normal	(300-1,000 ng/dL)
RFT	Unremarkable	-
LFT	Unremarkable	-

over the scalp associated with headache and irritation. The swelling was skin colored and was not tender nor itchy. Over the period of multiple months, the swelling became diffused with increased severity leading to folds formation and hair loss. The patient did not seek any medical advice regarding this swelling, until 2 months ago when he was presented to the clinic, because of continuous bullying at school. The patient denied any ophthalmological, cardiac or other systems complaints.

On examination: A soft diffused swelling covering the back of the scalp was noticed. There were 7 folds and furrows running in an anteroposterior direction on the parietal and occipital areas of the scalp. There were oily scales between the

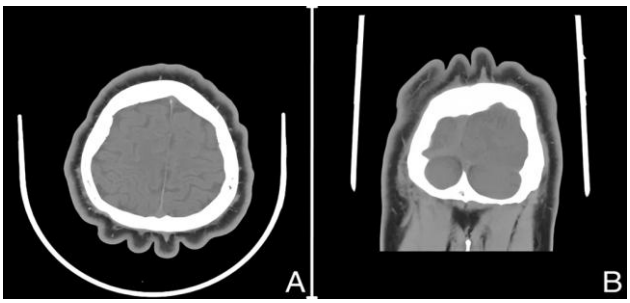

Figure 2. CT scan axial (A) and coronal (B) views showing thinking of the scalp with ridges and furrows involving the dermis and subcutis in the parietal and occipital regions (Reprinted with permission of patient)

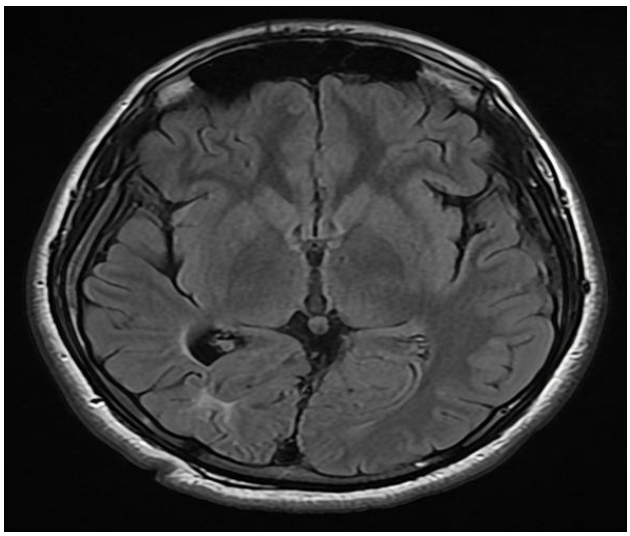

Figure 3. MRI scan T2-FLAIR axial view showing hyperintense subcortical white matter lesion involving the right occipital lobe associated with volume loss in the occipito-parietal lobe and posterior temporal region (Reprinted with permission of patient)

folds associated with bad odor. Lesions are not flattened by direct pressure or traction (**Figure 1**).

Laboratory investigations include full blood count, HIV, hepatitis serology, prolactin, T3, T4, TSH, and growth hormone. Vitamin D, testosterone, renal function test, and liver function test were ordered for the patient to exclude secondary causes (**Table 1**). The patient underwent computed topography to exclude any space-occupying lesion. The scan demonstrated thickening of the scalp (parietal and occipital region) with ridges and furrows involving dermis and subcutis resembling the surface of the cerebral cortex with no underlying lipoma. (**Figure 2**). The first impression was CVG.

Magnetic resonance imaging of the brain showed volume loss involving the right occipito-parietal lobes and posterior aspect of the right temporal lobe with subcortical abnormal white matter of high T2-FLAIR signal intensity, ex vacuo dilation of adjacent lateral ventricle. No significant interval changes. Abnormality involving the right cerebral hemisphere suggestive of old insult (**Figure 3**).

An electroencephalogram was done for the patient and showed abnormal intermittent generalized slow waves in addition to intermittent focal flow which may indicate mild non-specific encephalopathy. A biopsy of scalp was performed.

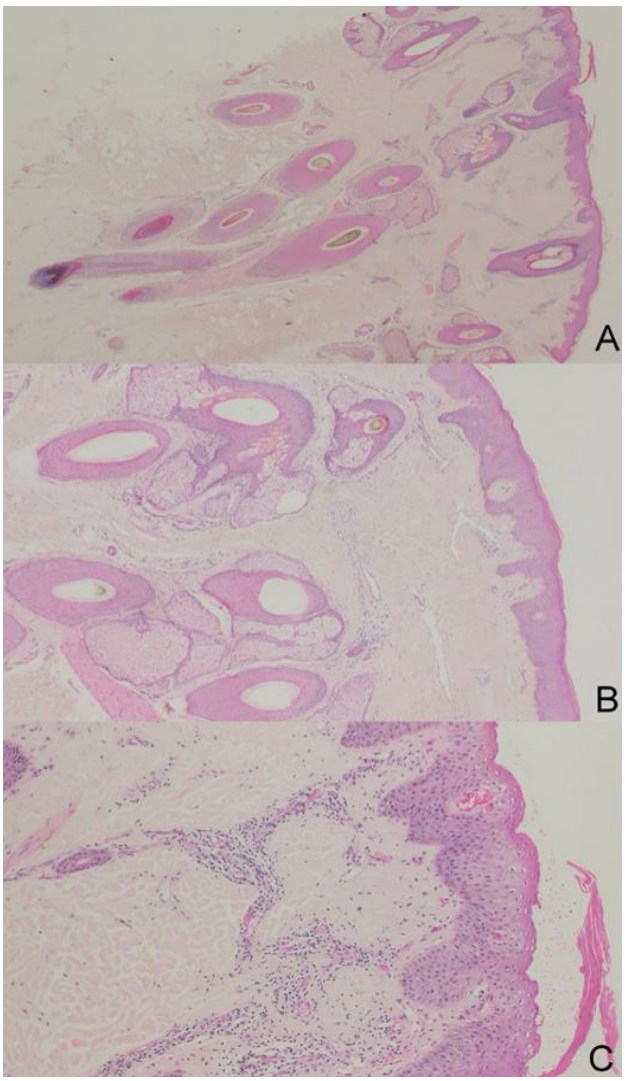

Figure 4. (A) Microphotograph showing skin punch biopsy from the epidermis to subcutis showing hypertrophy and hyperplasia of the adnexal structures (H&E, 20x), (B) Microphotograph showing hypertrophy and hyperplasia of the adnexal structures with mild perivascular lymphocytic inflammation (H&E, 100x), & (C) Microphotograph showing focal parakeratosis with increase in collagen fibers and mild to moderate perivascular lymphocytic inflammation (H&E, 200x) (Reprinted with permission of patient)

Histopathological examination revealed stratified squamous epithelium with focal parakeratosis along with hypertrophy and hyperplasia of adnexal structure, increased in collagen fibers and mild perivascular lymphatic inflammation. Features compatible with CVG (**Figure 4**).

DISCUSSION

Few case reports and studies have documented the coexistence of CVG and epilepsy. In a case study published in the Indian Journal of Dermatology, a 39-year-old man presented with CVG and a long-standing history of epilepsy [5]. Another study published in the Journal of Clinical Neuroscience described a case of CVG associated with focal epilepsy in a 30-year-old male [6]. These reports suggest that there may be a shared pathogenic mechanism or genetic predisposition that contributes to both conditions.

The exact relationship between CVG and epilepsy remains unclear, and further research is needed to elucidate the underlying mechanisms. However, some hypotheses have been proposed. One theory suggests that the abnormal folding and thickening of the scalp in CVG may exert pressure on the underlying brain tissue, leading to disturbances in electrical activity and potentially triggering seizures [4].

Another hypothesis suggests that there may be common genetic factors or signaling pathways involved in the development of both CVG and epilepsy [7]. In a 2016 study published in the American Journal of Medical Genetics [8], the analysis of 62 cases of CVG revealed a consistent correlation between CVG and significant psychomotor delay. The majority of patients exhibited an inability to walk or talk, and a significant portion experienced difficulties in performing activities of daily living. It is important to highlight, however, that our patient's case deviates from this observed pattern. Contrary to the typical presentation, our patient demonstrated the ability to successfully engage in daily activities and achieve psychomotor milestones without notable delays.

In a second study published in a clinical case reports journal in 2020, two cases of drug-resistant epilepsy with CVG were evaluated [9]. Previous reports have indicated that conditions like acromegaly and low testosterone levels can potentially lead to CVG. A study from 1964 even reported that castration resolved CVG in two cases [10]. However, our patient's case contradicts these findings, as they had normal growth hormone and testosterone levels.

In contrast to previous case reports where individuals with CVG and epilepsy typically exhibit normal brain magnetic resonance imaging (MRI) results, our case presentation reveals an abnormality in the patient's MRI. This abnormality suggests a previous injury, which could be one of the contributing factors to the patient's current condition. The presence of encephalomalacia indicates the possibility of an ischemic stroke that occurred during the prenatal, natal, or childhood period which may have led to the current situation.

The early onset of CVG which preceded the development of epilepsy in our case, demonstrating the possibility of having CVG as an early indicator of epilepsy or other neurological conditions. Numerous case reports have examined potential associations and causal factors related to CVG. For instance, a study documented a case in which secondary CVG developed in a 46-year-old female patient with cerebriform intradermal nevus [11].

Furthermore, an Italian article published in 2022 reported two cases of CVG occurring in patients with Noonan syndrome [12]. Additionally, a recent case report published in 2022 highlighted a case of CVG presenting in a patient diagnosed with synovitis, acne, pustulosis, hyperostosis, and osteitis [13]. These reports contribute to the growing body of literature exploring the diverse etiologies and manifestations of CVG. As evident from previous reports, the coexistence of epilepsy and CVG can occur even in the absence of typical hypothetical causes. It is crucial to remain open to new and innovative explanations for this potential relationship. Although certain characteristics, such as male gender and early onset, are commonly observed among affected patients, the underlying origins of this association are still unclear. Furthermore, physicians should be aware that CVG could be an early indicator of epilepsy or other neurological disorders.

CONCLUSION

Based on clinical features, laboratory findings, and imaging results, a diagnosis of CVG is made. The patient was instructed to follow up with neurology and dermatology in addition to the possibility of surgical referrals when needed with plastic surgery.

Also, the patient was referred to consult the psychiatrist to assess his personality changes. Proper skin care education and neurological evaluation have been made with clear instructions.

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Data sharing statement: Data supporting the findings and conclusions are available upon request from the corresponding author.

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