Cutis Verticis Gyrata and epilepsy, is there a typical patient?

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April 04, 2024

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No conflict of interest to be reported.

No funding for this article.

Patient Consent have been Obtained.

Key words:

Epilepsy, Cutis Verticis Gyrata, Neurology, Dermatology.

Key Clinical Message:

Epilepsy and CVG can coexist even without typical known causes. It's important to be open to new explanations for this relationship. While certain characteristics 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.

Introduction:

Cutis verticis gyrata (CVG) is a rare dermatological condition characterized by thickening and folding of the scalp, resulting in a corrugated or ridged appearance. The term "cutis verticis gyrata" translates to "wrinkled skin of the scalp," and it typically affects the vertex or top of the head. This condition is often congenital, but it can also develop later in life due to factors such as trauma, inflammatory conditions, or tumors (1). The folds and ridges in CVG are caused by an increase in skin and underlying tissue, including the dermis and subcutaneous fat (2).

CVG is occasionally associated with other medical conditions, including neurological disorders such as epilepsy. 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 History/examination:

A 15 years old Saudi male, right handed with unremarkable family history. He is a son of a 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 PICU admission due to respiratory distress. 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 out of stock and he was shifted to levetiracetam and carbamazepine in 2021. During these years, the patient had episodes of visual hallucinations and dizziness which resolved after the 3rd year of treatment. Afterwards, the patient showed great improvement and the last documented convulsion was 3 years ago, so he was prescribed only levetiracetam once daily.

One year before the patient was diagnosed with epilepsy, he complained of diffused swelling over the scalp associated with headache and irritation. The swelling was non tender, non itchy and skin colored. Over the period of multiple months, swelling has become more diffused and severe to the extent it caused folds formation and hair loss. Patient did not seek any medical advice regarding this swelling, until 2 months ago he presented to our clinic, as a result of continuous bullying at school. Patient denied any ophthalmological or cardiac complain.

On examination: A soft diffused swelling covering the back of the scalp was noticed. There were folds (7) and furrows running in an anteroposterior direction on the parietal and occipital areas of the scalp. There were oily scales between the folds associated with bad odor. Lesions are not flattened by direct pressure or traction.

Methods (differential diagnosis, investigations and treatment):

Labs:

CBC:

WBC: Normal

Hgb: High (18.2 g/dl)

MCV: Low (75.7 FL)

RDW-CV: High (16.3%)

Lymphocyte: High (58.4%)

Hepatitis Serology: -ve

HIV: -ve

Prolactin, T3, T4 and TSH: Normal.

Growth hormone: Normal.

Vitamin D: Low (15.8mg/ml)

Testosterone: Normal

RFT: Unremarkable

LFT: Unremarkable

CT of the Brain:

There is thickening of the scalp (Parietal and Occipital region) with ridges and furrows involving dermis and subcutis resembling the surface of the cerebral cortex. No underlying lipoma seen. Impression: Cutis Verticis Gyrata.

MRI of the Brain:

Volume loss involves 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.

Impression: No significant interval changes. Abnormality involving right cerebral hemisphere suggestive of old insult.

EEG:

Abnormal intermittent generalized slow in addition for intermittent focal flow wich may indicate mild encephalopathy non specific.

Biopsy:

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 Cutis Verticis Gyrata.

Conclusion and Results:

Based on the clinical features, laboratory findings, and imaging results, a diagnosis of Cutis Verticis Gyrata is made. The patient was instructed to follow up in Neurology and Dermatology with the possibility of surgical referral when needed. Proper skin care education and neurological evaluation have been made with clear instructions.

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 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 lead to the current situation.

The early onset of CVG which precedes the development of epilepsy in our case, demonstrates 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 Cutis Verticis Gyrata. 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 SAPHO (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.

References:

1. Garg T, Chander R, Vaidya V, et al. Cutis Verticis Gyrata: A Rare Case Report. Int J Trichology. 2010;2(2):116-118. doi:10.4103/0974-7753.77515

2. Alper M, Selden M, Weil R. Scalp Reconstruction for Cutis Verticis Gyrata Using a Superiorly Based Galeal Frontalis Muscle Flap. J Neurosurg. 2005;103(2):348-350. doi:10.3171/jns.2005.103.2.0348

3. Fisher RS, Acevedo C, Arzimanoglou A, et al. ILAE Official Report: A Practical Clinical Definition of Epilepsy. Epilepsia. 2014;55(4):475-482. doi:10.1111/epi.12550

4. Herskovitz I, Mutasim DF. Cutis Verticis Gyrata: A Review. J Eur Acad Dermatol Venereol. 2015;29(5):842-846. doi:10.1111/jdv.12532

5. Sathyanarayana B D, Archana M S, et al. Cutis Verticis Gyrata. Indian JDermatol. 2015;60(3):324. doi:10.4103/0019-5154.156388

6. Sinha S, Sachdeva N, Maheshwari MC. Cutis Verticis Gyrata with Focal Epilepsy. J Clin Neurosci. 2002;9(3):337-339. doi:10.1054/jocn.2001.1089

7. Goto M, Muro Y, Hatakeyama T, et al. Cutis Verticis Gyrata and Epilepsy with a Ring Chromosome 7. J Am Acad Dermatol. 2000;42(6):1057-1058. doi:10.1067/mjd.2000.104156

8. Tucci A, Pezzani L, Scuvera G, et al. Is cutis verticis Gyrata-Intellectual Disability syndrome an underdiagnosed condition? A case report and review of 62 cases. Am J Med Genet A. 2017;173(3):638-646. doi:10.1002/ajmg.a.38054

9. Rattagan M, De Francesco M, Kriebaum A, Ferraro F, Major C, Sharma D, Ojeda A, Martinez O, Musto AE. Cutis verticis gyrata: Two cases associated with drug-resistant epilepsy. Clin Case Rep. 2020 May 18;8(8):1365-1368. doi:10.1002/ccr3.2814. PMID: 32884755; PMCID: PMC7455441.

10. Akesson HO. Cutis Verticis Gyrata and Mental Deficiency in Sweden. I. Epidemiologic and Clinical Aspects. Acta Med Scand. 1964 Jan;175:115-27. PMID: 14110633.

11. Fronek LF, Braunlich K, Farsi M, Miller RA. A Rare Case of Cutis Verticis Gyrata with Underlying Cerebriform Intradermal Nevus. Cureus. 2019;11(12):e6499. doi:10.7759/cureus.6499

12. Mercadante F, Piro E, Busè M, et al. Cutis verticis gyrata and Noonan syndrome: report of two cases with pathogenetic variant in SOS1 gene. Ital J Pediatr. 2022;48(1):152. doi:10.1186/s13052-022-01340-4

13. Wang Y, Wang S, Zheng L, et al. Synovitis, Acne, Pustulosis, Hyperostosis, and Osteitis (SAPHO) Syndrome with Cutis Verticis Gyrata: Case Report and Review of Literature. Clin Cosmet Investig Dermatol. 2022;15:1415-1420. doi:10.2147/CCID.S372522

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