Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

A Case Report of a Cutis Verticis Gyrata-Intellectual Disability

K. Elhadri[1*], M.Raboua[1], F. Amenzouy[1], B .Boutaqiout[1], M. Idrissi Ouali[1], N. Cherif Idrissi Ganouni[1]

[1]Radiology Department Ar-Razi Hospital, Chu Mohammed VI University Cadi Ayad Marrakech, Morocco

DOI: 10.36347/sjmcr.2023.v11i05.020 | **Received:** 24.03.2023 | **Accepted:** 28.04.2023 | **Published:** 06.05.2023

***Corresponding author:** K. Elhadri
Radiology Department Ar-Razi Hospital, Chu Mohammed VI University Cadi Ayad Marrakech, Morocco

Abstract **Case Report**

Cutis Verticis Gyrata-Intellectual Disability (CVG-ID) syndrome is a rare neurocutaneous syndrome characterized by intellectual disability and scalp folds, furrows that are typically absent at birth and are first noticed after puberty. First reported in 1893, the syndrome was mainly identified in subjects living in psychiatric institutions. Most patients were reported in the literature during the first half of the 20th century. CVG-ID is now a less reported and possibly under-recognized syndrome. Here, we report a patient with CVG-ID that was diagnosed using the novel approach of magnetic resonance imaging.

Keywords: Intellectual disability; Cutis Verticis Gyrata epilepsy; brain MRI**.**

INTRODUCTION

Cutis Verticis Gyrata (CVG) is an abnormality of the scalp characterized by the formation of skin folds and furrows, that cannot be corrected by pressure or traction on the scalp. This abnormality was first described in 1837 by Alibert and has been reported since both as an isolated form or in association with other conditions.

The association of CVG with intellectual disability (ID) was first reported by Mc Dowell and Cowan in 1893, in a patient affected by severe ID and epilepsy with muscle wasting and contractures. All published patients with CVG were studied in 1953 by Poland and Butterworth, who classified CVG into primary and secondary forms.

Cutis verticis gyrata is a rare neurocutaneous disorder characterized by cutis verticis gyrata (CVG) and polymicrogyria, which is a condition where the surface of the brain has an excessive number of small folds or gyri. MRI is an important tool for the diagnosis and management of this condition.

CASE REPORT

A 32-year-old male patient, was born after regular gestation and delivery from non-consanguineous parents. His family history was unremarkable. At the age of 15 year, the patient experienced generalized tonic-clonic seizures; from that time on, a progressive intellectual regression was observed. During the clinical examination, an appearance of a longitudinal cutis verticis gyrata of the scalp was noted (figure 1A). Routine blood tests, electrocardiography and chest X-ray were all normal.

A brain MRI was performed and showed a longitudinal CVG with thickening of the skin and subcutaneous tissues (figure 1B,C), bilateral polymicrogyria involving more marked in the occipital lobe associated with colpocephaly, atrophy of the corpus callosum (figure 2). We also found hyper intensity with left frontoparietal cortical dysplasia.

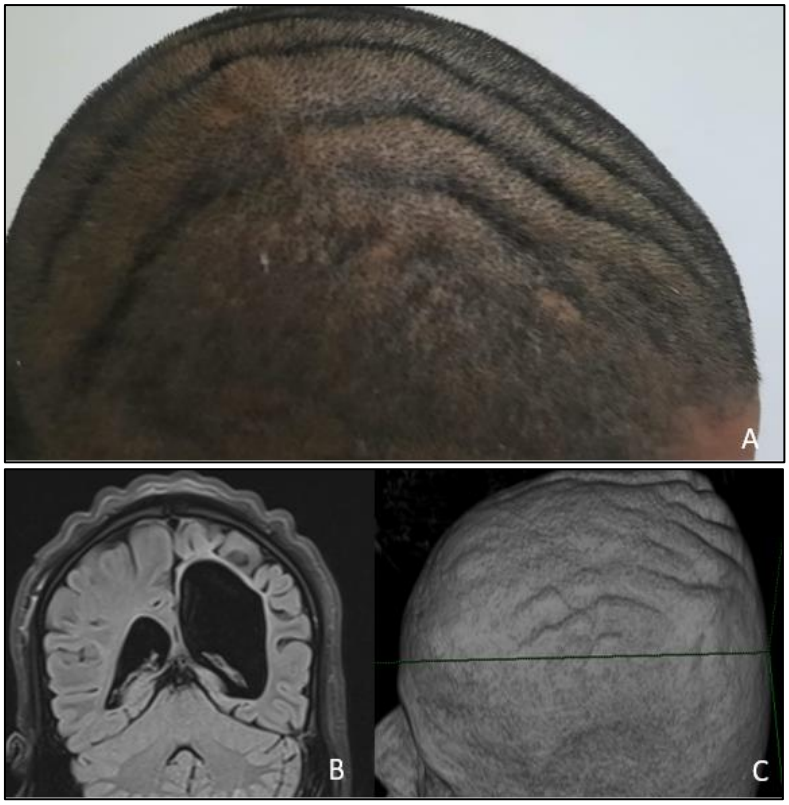

Figure 1: A: clinical photography showing anteroposteriorly oriented ridges of the scalp that simulates surface of the brain, that cannot be corrected by pressure or traction on the scalp. B: coronal FLAIR demonstrating longitudinal CVG with thickening of the skin and subcutaneous tissues. C: Three-dimensional magnetic resonance imaging (3D MRI) showing typical scalp folds.

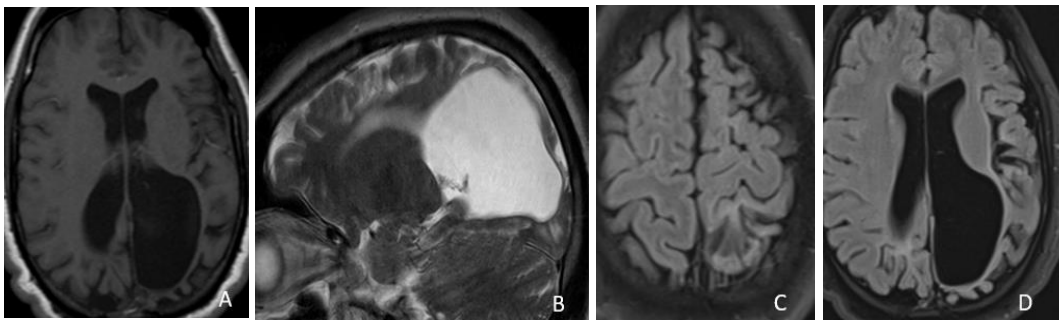

Figure 2 : axials T1 (A), FLAIR (C, D) and coronal T2 weighted magnetic imaging showing :
-Bilateral polymicrogyria involving more marked in the occipital lobe associated with colpocephaly.
-Hyperintensity and left frontoparietal cortical dysplasia.

DISCUSSION

Cutis verticis gyrata is an uncommon condition resulting from the abnormal growth and folding of the scalp, leading to the formation of ridges and furrows resembling the gyri of the brain. Its estimated incidence is 1 in 100,000 in males and 0.026 in 100,000 in females [1]. Some authors suggest that the higher incidence in men may be due to easier detection resulting from shorter haircuts [2]. Additionally, a higher prevalence of CVG was found among psychiatric patients in institutionalized settings (0.71-3.4%) compared to the general population [3].

The etiology of CVG-ID remains unknown, which has resulted in the condition being largely forgotten and poorly understood, although there is evidence for both endocrinologic dysfunction and a genetic etiology [4]. Cutis verticis gyrata is classified into primary (idiopathic) and secondary forms. The primary form usually develops after puberty and the folds tend to be symmetric. The primary CVG is further divided into essential and nonessential types. The essential primary form of CVG is not associated with any underlying medical conditions. In contrast, the nonessential form, as seen in our case, is associated with mental deficiency, epilepsy, seizures, various ophthalmic abnormalities, and is considered a neurocutaneous syndrome [3]. Secondary CVG can result from various underlying medical conditions, such as acromegaly, scalp dermatologic conditions

(including benign or malignant tumors, nevi, inflammatory dermatoses, trauma), Type 2 diabetes, hypothyroidism, and amyloidosis, all of which have been associated with CVG [1, 2, 3]. Additionally, several syndromes have been reported in association with CVG, including Ehlars-Danols syndrome, Turner syndrome, Beare-Stevenson syndrome, Apert syndrome, Noonan syndrome, Klinefelter syndrome, and Lennox Gastaut syndrome [4, 5].

In our case, the direction of CVG folds is anterior-posterior (longitudinal), although it may also be transverse in the vertex and occipital regions [7]. On MRI, the coronal plane is the best plane to visualize the longitudinal folds in the anterior-posterior direction (Figure 1B), while the sagittal plane is the best for the transverse folds [7]. Three-dimensional MRI with surface rendering has been found to be very useful in demonstrating the scalp folds of CVG (Figure 1C) [9]. The identification of CVG on MRI studies has important diagnostic implications and underscores the need for clinicians to be aware of this condition. It is important to note that CVG may be asymptomatic and may not be visible on physical examination, making MRI a useful tool for identifying this condition.

Brain anomalies detected by computed tomography (CT) or MRI are found in about 38% of patients. Cortical or subcortical atrophy is the most common finding and has been described in all brain areas including occipital, parietal, and in the posterior fossa structure. Occasional findings include polymicrogyria [10], microcephaly, colpocephaly of the occipital horns, and pronounced cerebellar folia with small splenium [11] or extensive periventricular calcifications [12].

The abnormalities found in our MRI case involve bilateral polymicrogyria, more pronounced in the occipital lobes. We observed a subcortical signal abnormality that extended to the surface of the ventricles, which showed iso-signal on the T1 sequence and hyper-signal on the T2 and FLAIR sequences, but not on the diffusion sequence, indicating cortical dysplasia. This subcortical signal abnormality produced a transmantle sign. Additionally, we also observed dedifferentiation of grey and white matter and splenium callosum dysgenesis associated with colpocephaly.

CONCLUSION

In conclusion, this case report highlights the importance of considering the diagnosis of CVG-ID in patients with CVG, mental retardation, and epilepsy. MRI can be a useful tool for identifying CVG and associated neurocutaneous disorders. Further research is needed to fully elucidate the etiology of CVG and its relationship to underlying pathology.

REFERENCES

1. Yang, J. J. H., Sano, D. T., Martins, S. R., Tebcherani, A. J., & Sanchez, A. P. G. (2014). Primary essential cutis verticis gyrata-case report. *Anais brasileiros de dermatologia*, *89*, 326-328.
2. Snyder, M. C., Johnson, P. J., & Hollins, R. R. (2002). Congenital primary cutis verticis gyrata. *Plastic and reconstructive surgery*, *110*(3), 818-821.
3. Chang, G. Y. (1996). Cutis verticis gyrata, underrecognized neurocutaneous syndrome. *Neurology*, *47*(2), 573-575.
4. Fox, L. P., Geyer, A. S., Anyane-Yeboa, K., & Garzon, M. C. (2005). Cutis verticis gyrata in a patient with Noonan syndrome. *Pediatric dermatology*, *22*(2), 142-146.
5. Keller, K., Williams, C., & Seagle, B. (2001). Klinefelter syndrome and cutis verticis gyrata. *American Journal of medical genetics*, *103*(3), 249-251.
6. Chang, G. Y. (1996). Cutis verticis gyrata, underrecognized neurocutaneous syndrome. *Neurology*, *47*(2), 573-575.
7. Okamoto, K., Ito, J., Tokiguchi, S., Ishikawa, K., Furusawa, T., & Sakai, K. (2001). MRI in essential primary cutis verticis gyrata. *Neuroradiology*, *43*, 841-844.
8. Oh, D. J., Park, J. H., Kang, S. H., Hwang, S. W., & Park, S. W. (2006). Primary non-essential cutis verticis gyrata revealed with 3-D magnetic resonance imaging. *Acta dermato-venereologica*, *86*(5), 458-459.
9. Oh, D. J., Park, J. H., Kang, S. H., Hwang, S. W., & Park, S. W. (2006). Primary non-essential cutis verticis gyrata revealed with 3-D magnetic resonance imaging. *Acta dermato-venereologica*, *86*(5), 458-459.
10. Striano, S., Ruosi, P., Guzzetta, V., Perone, L., Manto, A., & Cirillo, S. (1996). Cutis Verticis Gyrata—Mental Deficiency Syndrome: A Patient with Drug-Resistant Epilepsy and Polymicrogyria. *Epilepsia*, *37*(3), 284-286.
11. Farah, S., Farag, T., Sabry, M. A., Simeonov, S. T., Al-Khattam, S., Abulhassan, S. J., ... & Al-Awadi, S. A. (1998). Cutis verticis gyrata-mental deficiency syndrome: report of a case with unusual neuroradiological findings. *Clinical dysmorphology*, *7*(2), 131-134.
12. Vanhoenacker, F. M., De Grave, C., & Catry, F. (2007, October). Cutis verticis gyrata associated with periventricular calcifications. In *RöFo-Fortschritte auf dem Gebiet der Röntgenstrahlen und der bildgebenden Verfahren* (Vol. 179, No. 10, pp. 1074-1075). © Georg Thieme Verlag KG Stuttgart· New York.