Cutis verticis gyrata = 頭皮の蛇様な変化

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Cutis verticis gyrata (CVG) is a descriptive term for a condition of the scalp manifesting as convoluted folds and furrows formed from thickened skin of the scalp resembling cerebriform pattern. Although Alibert first mentioned it, Robert described the condition in 1843. Unna introduced the term cutis verticis gyrata in 1907. Polan and Butterworth established the classification of cutis verticis gyrata in 1953, dividing cutis verticis gyrata into primary and secondary forms. In 1984, Garden and Robinson improved the classification by proposing new terms: primary essential cutis verticis gyrata for cases in which no other abnormality was found (rare) and primary nonessential, which can be associated with mental deficiency, cerebral palsy, epilepsy, schizophrenia, cranial abnormalities (microcephaly), deafness, ophthalmologic abnormalities (cataract, strabismus, blindness, retinitis pigmentosa), or a combination of these.

Secondary cases of cutis verticis gyrata are associated with the following underlying diseases:

- Pachydermoperiostosis
- Melanocytic nevi or hamartomas (cerebriform intradermal nevus)
- Neurofibroma
- Fibroma
- Dermatofibroma
- Cylindroma
- Nevus lipomatosus
- Connective tissue nevus
- Acromegaly
- Pseudoacromegaly
- Diabetes mellitus
- Autosomal dominant insulin-resistant syndrome
- Myxedema
- Graves disease
- Cretinism
- Amyloidosis
- Posttraumatic (eg, traction alopecia)
- Inflammatory processes (eg, eczema, psoriasis, Darier disease, folliculitis, impetigo, erysipelas, atopic dermatitis, acne conglobata)
- Syphilis
- Focal mucinosis
- Leukemia
- Fallopian tube carcinoma
- Acanthosis nigricans
- Beare-Stevenson syndrome
- Tuberous sclerosis
- Ehlers-Danlos syndrome
- Noonan syndrome
- Turner syndrome
- Supernumerary X chromosome syndromes (including Klinefelter syndrome)
- Hereditary neuralgic amyotrophy
- Intracranial aneurysm
- Intraventricular ependymoma
- Hyperimmunoglobulin E syndrome

Pathophysiology

In the primary essential form, the etiology is not known, and, though most of the cases seem sporadic, autosomal recessive and autosomal dominant inheritance with variable expression have been described. In the primary nonessential form, the pathogenesis (beside the genetic determination) may have an endocrinologic basis.

Cutis verticis gyrata mainly occurs in males, after puberty, and it may disappear after castration. This may be due to increased peripheral use of testosterone, which was further supported by the results of the study in which the free testosterone level was reduced in patients with primary cutis verticis gyrata compared with controls.

Male predominance may also suggest an X-linked inheritance. An association with the fragile X syndrome or other fragile sites on chromosomes 9, 10, and 12, and, in a single case, breaks at bands 3p14 and 16q23, has been reported.

In the secondary form, the etiology depends on the underlying process (eg, inflammatory,
neoplastic). Lymphedema is a postulated cause of cutis verticis gyrata in Turner and Noonan syndromes

History

In primary cutis verticis gyrata, skin plaques develop after puberty, usually in the vertex and occipital region. In secondary cutis verticis gyrata, skin plaques can be present at birth. The skin gradually becomes thicker, and folds and furrows are formed.

No other symptoms are usually present; however, pain was observed in cases with cutis verticis gyrata due to intradermal nevus and in traction alopecia. The progression of changes is visible.

Physical

In the primary form, only the scalp is involved. Cutis verticis gyrata typically affects the vertex and occipital region, but some forms can involve the entire scalp. Folds are soft and spongy and cannot be corrected by pressure or traction. In primary cutis verticis gyrata, folds are usually symmetric; in secondary cutis verticis gyrata, folds are asymmetric.

In most cases, the direction of the folds is anterior to posterior, but it may be transverse in the occipital region. The number of folds varies from 2-12, although some atypical cases with 1 fold have been described. The skin color is unchanged. The hair over the folds may be sparse but normal in the furrows. Maceration and an unpleasant smell may be present in patients with secondary infections in the furrows.

Causes

The cause is unknown in primary cases, although genetic and endocrinologic factors are suspected to participate in the etiology. Systemic diseases, inflammatory dermatoses, underlying nevoid abnormalities, and trauma are most common in secondary cases.

Laboratory Studies
In primary cases, no laboratory tests should be ordered. A low free testosterone level was observed in some patients with primary cutis verticis gyrata.\(^{30}\)

In secondary cases, laboratory tests depend on the presentation and the associated disease.

### Imaging Studies

Perform magnetic resonance imaging (MRI) or computed tomography (CT) in cutis verticis gyrata presenting at birth or when associated with mental retardation and neurologic and/or ophthalmologic abnormalities to determine or exclude any structural brain abnormalities.\(^{45,46,47}\)

Some authors suggest a standard anteroposterior skull film with soft-tissue technique, which may be helpful for making the diagnosis in subtle cases.

In CT scanning, thickening of the skin and subcutaneous fat and irregularly distributed cutaneous folds can be observed. Changes, such as cortical-subcortical atrophy, a dilated ventricular system, abnormal brain calcifications, bone changes, or an intracranial tumor, were also demonstrated in patients with cutis verticis gyrata. In patients with acromegaly and cutis verticis gyrata, enlarged sellae with enhancing intrasellar masses were present.\(^{11,48,49}\)

In MRI, severe abnormality of the occipital lobes, bilateral polymicrogyria, small frontal and anterior temporal lobes, parietal and occipital cortex atrophy, colpocephaly, hypoplastic splenium of the corpus callosum, and atrophy of the cerebellar cortex were described.\(^{50,51}\)

In essential primary cutis verticis gyrata, MRIs reveal thickened dermis and a slight increase in the volume of subcutaneous fat.\(^{46,52}\)

### Other Tests

Always obtain a skin biopsy specimen of the affected area of the scalp to identify the etiology of cutis verticis gyrata.\(^{8,9}\)

Evaluate female infants with cutis verticis gyrata with or without peripheral lymphedema by chromosomal analysis to exclude Turner syndrome.

Obtain an EEG in all patients with mental deficiency and cutis verticis gyrata to establish the nature of the mental disorder. Among others, severe diffuse slowing of the background activity...
with very frequent right temporal spikes and spike-wave complexes that spread to the frontal area were described.\textsuperscript{53}

**Histologic Findings**

The histopathologic picture is of normal appearance in most cases of the primary type. In some cases, dermal collagen thickening, hypertrophy of the pilosebaceous structures, and multiple sweat glands and ducts may be present.\textsuperscript{54} In the secondary form, the histopathologic picture depends on the underlying disease.

**Medical Care**

Properly establishing the diagnosis is very important. Separately exclude or treat any underlying process (see Background). Primary essential cutis verticis gyrata is a cosmetic problem, but psychological repercussions are important.

Hygiene for folds and furrows is very important. In some patients, using medicated shampoos may be beneficial.

**Surgical Care**

In primary cutis verticis gyrata, surgical resection of the lesions is usually requested for psychological or esthetic reasons.\textsuperscript{54,29,55,56,57,58,59}

In cases of cerebriform intradermal nevus, early diagnosis, wide surgical excision, and plastic reconstruction should be always considered.\textsuperscript{7,8,44}

**Consultations**

Consult a surgeon to establish the possibility of surgical excision. Consider a consultation with a neurologist if any suspicion exists of an underlying neurologic process. Additionally, consider a consultation with an ophthalmologist if any suspicion exists of an underlying ophthalmologic abnormality.