CASE REPORT

Essential primary cutis verticis gyrata

Leticia K. Schenato¹, Tatiane Gil¹, Lauro A. Carvalho¹, Nelson Ricachnevsky²,
Alberto Sanseverino³, Ricardo Halpern⁴

Abstract

Objective: to review the indications, main steps and complications of bone marrow transplantation in children.

Sources: Medline-based literature review.

Summary of the findings: we comment about the indications of autologous, allogeneic and syngeneic bone marrow transplantation, donor selections, harvest and infusion of the hematopoietic progenitor cells that will reconstitute the hematopoietic and immune systems. We describe the different conditioning regimens and the new sources of cells, such as cord blood. We also describe the most common events after the procedure, including infections, graft versus host disease, and cardiovascular, pulmonary, hepatic, genitourinary, and gastrointestinal complications. The late effects and their impact on quality of life are also discussed.

Conclusions: bone marrow transplantation does not confer an absolutely normal life span to all the patients; however, it represents the only chance of cure for children with certain neoplastic or immunological diseases. By knowing the steps of the procedure, pediatricians can be a source of information on bone marrow transplantation to the patients and their families.


Introduction

Cutis verticis gyrata (CVG) is characterized by excessive growth of the scalp skin, leading to the formation of furrows that resemble cerebriform pattern.¹⁻³ It was first mentioned by Alibert in 1837; however Robert described the condition in 1843.⁴ Unna introduced the term Cutis verticis gyrata, which is used until today, in 1907.⁴

CVG is classified into two forms: primary (essential and nonessential) and secondary.⁴ The primary nonessential form affects 0.5% of mentally retarded patients; cerebral palsy, epilepsy, cataract, and blindness may be observed.¹,⁵ The primary essential form is not associated with neurological and ophthalmologic abnormalities, usually occurring at the pubertal stage, exclusively in men; however, inheritance has not been confirmed.¹,⁴,⁵ In primary essential CVG, cerebriform folds are formed on the scalp. The secondary forms of CVG often include pachydermoperiostosis, acromegaly and intradermal cerebriform nevus.¹

In children, the cases of CVG, regardless of their etiology, are extremely rare. Nevertheless, the primary nonessential form,⁶,⁷ association with genetic syndromes⁸,⁹ and familial cases¹⁰ are reported in the literature (Table 1).

There are only two reports of primary essential CVG in the literature worldwide. These two cases consisted of male
Table 1 - Etiology of Cutis verticis gyrata in children

<table>
<thead>
<tr>
<th>Etiology of CVG</th>
<th>Comments on the case</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>Beare-Stevenson syndrome</td>
<td>Newborn infant with CVG by early fusion of skull sutures, causing great deformity of the calvarium. Associated with other malformations such as: syndactyly, ankylosis and synostosis of the hands, feet, and spine, and different levels of mental retardation.</td>
<td>8</td>
</tr>
<tr>
<td>Intradermal nevus</td>
<td>Newborn with Noonan syndrome, involvement of multiple organs, chylothorax, and CVG by intradermal nevus. This is the only case in which CVG and Noonan syndrome are associated.</td>
<td>9</td>
</tr>
<tr>
<td>Primary nonessential form</td>
<td>Newborn with CVG, neurological deficit, and other slighter congenital malformations; normal skin biopsy; persistent hypotonia in the 7th month and remarkable growth delay. This is the only case of a newborn infant with this type of CVG. Case of two brothers with CVG, microcephaly, retinitis pigmentosa cataract, sensorineural deafness, and mental retardation.</td>
<td>6 and 7</td>
</tr>
<tr>
<td>Family form</td>
<td>Family form of CVG, in which mother and child had one large scalp skinfold, without any other associated disease. This is the only case of the family form of CVG.</td>
<td>10</td>
</tr>
</tbody>
</table>

adult patients without underlying neurological or ophthalmologic abnormalities, in which possible causes of secondary CVG were ruled out. There is no case of primary essential CVG described in the literature worldwide.

In this case report, we describe the rare case of a male child with primary essential CVG, including diagnostic and therapeutic aspects.

Case report

Nine-year-old, non-white male, from the state of Santa Catarina, presented with 27-cm² scalp lesions resembling cerebriform pattern. The lesions affected the parietal and occipital regions on both sides, especially the left-handed side, and were soft and doughnut-shaped, with hair growth (Figures 1 and 2). Three years ago, the patient had developed a 5cm² lesion in the occipital region, which evolved into gradual increase in fold size. Our patient was born to non-blood-related parents, and is their third and last child. The mother denies the presence of pigmented scalp lesions at birth. There is no family history of this condition. The child is now attending the 3rd grade in elementary school, but did the first grade twice; he started to walk at the age of one and a half years and began to speak at the age of two. Physical examination revealed no signs of acromegaly; his neurological and ophthalmologic exams did not show any abnormalities; his IQ was considered high.

Complementary exams

- **Lab exams**: hemogram, GOT, GPT, alkaline phosphatase, creatinine, urea, calcium, free T4 and TSH: within normal limits. Nonreactive VDRL.
- **Electroencephalogram**: scarce acute potentials in the left parietal region; adequate tracing according to age.
- **Cranial CT scan**: normal cranial contents, extracranial mass caused by tumefaction of soft tissues in parietal and occipital regions.
Discussion

CVG is characterized by excessive growth of the scalp skin with the formation of furrows towards the anteroposterior direction, resembling cerebriform pattern.\(^1\)-\(^3\)

Primary CVG is grouped into two forms\(^3\):

- **Nonessential**: presence of scalp cerebriform folds associated with neurological abnormalities; IQ is rarely greater than 35. Microcephaly, static encephalopathy and seizures may be observed; ophthalmologic abnormalities (cataract, strabismus and blindness) may also be detected.\(^1\),\(^3\),\(^5\),\(^12\),\(^13\)

- **Essential**: extremely rare form of thickened skin of the scalp resembling cerebriform pattern; more predominant in males, with no association with neurological and ophthalmologic diseases.\(^3\),\(^4\),\(^11\),\(^13\) In most cases, the onset of this type of CVG occurs during or after puberty, and in 90% of the patients, it appears after the age of 30.\(^1\),\(^4\),\(^5\),\(^13\) Histological findings include hypertrophy of sebaceous structures, and no evidence of collagen thickening\(^1\)-\(^3\); no malignant abnormality is observed in the skin or in the brain parenchyma.\(^5\),\(^12\)

In the medical literature worldwide, there are only two reports of primary essential CVG. The first case was described by Garden\(^4\) in a 26-year-old patient, who noted the formation of small folds on the scalp and, after four years, found a large CVG lesion in parietal regions; the patient complained of pressure in the head, mild itching, and thinning of hair; there was neither family history of a similar disease nor history of consanguineous marriage; clinical examination showed no abnormalities; lab exams for the detection of endocrine diseases yielded negative results. The second case was reported by Cribier\(^11\) in a 19-year-old patient, who noted moderate thickening of the scalp at the age of 15 and progressively developed folds in the occipital and parietal regions; the hair had normal appearance; the comprehensive neurological exam carried out did not show any abnormalities; the patient’s IQ was normal; x-ray and CT scan of the brain did not show bone involvement; the patient underwent a surgery for scalp reduction.

Our patient developed scalp folds at the age of six, with gradual increase in size, during three years, until they reached the current size. This is different from other cases.
of primary CVG, in which lesions usually appear in adolescence or at the beginning of adult life. Clinical and complementary exams confirmed the absence of neurological and ophthalmologic abnormalities, and of bone involvement, allowing us to rule out secondary causes of CVG, such as pachydermoperiostosis and acromegaly (normal cranial CT), thyroid diseases (normal T4 and TSH) and syphilis (nonreactive VDRL). Skin biopsy was not performed because the family did not give their consent, since they considered the procedure a physical aggression. We did not regard biopsy as necessary for the diagnosis of primary essential CVG, since clinical history and lab exams helped us rule out other possible causes of CVG (intradermal cerebriform nevus, for example). Intradermal nevus is usually present at birth or develops early on in life, is predominant in females, and consists of gradual hair loss and increase of lesion size in adolescence.

After an extensive literature review, we did not find any case of primary essential CVG in children. There are few reports of CVG in children; among these, the most relevant cases include: CVG in Noonan syndrome; in Beare-Stevenson syndrome; familial form of CVG and due to the primary nonessential form (Table 1).

The differential diagnosis of primary essential CVG includes the nonessential form, pachydermoperiostosis, acromegaly, intradermal nevus and other less frequent causes of CVG:

- **Primary nonessential CVG**: may be associated with mental retardation and chronic schizophrenia. Schepis studied the prevalence of CVG in 494 patients from a psychiatric institution; 22 patients (21 men) had primary CVG. Another study was conducted on the presence of CVG in 83 hospitalized psychiatric patients; 3 men had CVG. This high prevalence rate in men with psychiatric disorders may be explained by ethnical factors or by the habit of shaving off the hair of these patients, which facilitates diagnosis.

---

**Figure 2** - CVG lesion covering great part of the scalp, especially the left parietal and occipital region
- **Pachydermoperiostosis**: occurs by clubbing of the toes, periostal bone formation, thickening of facial skin, with enhancement of furrows, lesions caused by hypertrophy of sebaceous structures, by CVG and by palmoplantar hyperhidrosis. The primary form is transmitted by autosomal dominant inheritance; skin and bone abnormalities progress severely during 5 to 10 years and, after that, they remain unchanged for the rest of life; several patients have mental retardation. The secondary form occurs in men aged between 30 and 70 years; bone alterations are very characteristic, develop very fast and may be painful; usually caused by severe pulmonary disease such as bronchial carcinoma.

- **Acromegaly**: characterized by excessive growth of facial bones and skull. The presence of CVG is not uncommon in cases of acromegaly; therefore, its presence should warn physicians against the possibility of hypophyseal adenoma; for this reason, we requested a cranial CT scan of our patient.

- **Intradermal nevus**: usually present at birth or very early during childhood under the form of a small hyperpigmented area, which gradually increases in size at the pubertal stage and may cover a considerable portion of the scalp; progressive alopecia is the general rule. The incidence is higher among females. In our case, no hyperchromic maculopapular lesions were reported at birth and the lesion began to gradually increase at the age of six and not close to the pubertal stage. The diagnosis of large, congenital intradermal nevus should be followed by complete surgical exeresis of the lesion, since it has a great potential to evolve into malignant melanoma.

- **Other medical conditions associated with CVG**: inflammatory diseases of the scalp (eczema, psoriasis, folliculitis, impetigo, erysipela and pemphigus), myxedema, leukemia, syphilis, acanthosis nigricans, tuberous sclerosis, Ehlers-Danlos syndrome, amyloidosis and diabetes mellitus.

The treatment of CVG varies from observation and care of the affected region to the surgical exeresis of excessive skin. In general, the affected area is asymptomatic; however, there may be deposits of secretions that cause unpleasant smell and itching. Therefore, cleaning the scalp regularly and properly helps alleviate the symptoms. Our patient did not show symptoms in the region affected by CVG, because his mother was very careful, cleaning the area and shaving off the hair to prevent the deposit of residues.

The esthetic aspect draws more attention due to the involvement of an extensive area. Different techniques for the reduction of scalp disorders are available for the treatment of CVG: total resection of the lesion and grafting; placement of a skin expander on the healthy area and grafting; partial resection of the most prominent portion of the lesion. Tissue expansion has allowed for good quality correction in areas where conventional flap and grafting techniques are an obstacle; one of the main advantages is the reduced number of surgeries; the disadvantages consist of esthetic deformity produced by the expander. In cases in which the involvement of the scalp is very large, partial resections of the most prominent lesions can be performed. When total resection of extensive lesions is desired, myocutaneous flaps or free flaps may be chosen; the latissimus dorsi muscle is currently selected for the treatment of extensive areas. However, the plastic surgical staff believed it was useless to submit the patient to a surgical procedure, since the total resection of the lesion would cause irreversible alopecia and the expansion of the skin in the healthy region would not be enough to cover all the affected area. Therefore, we did not use any surgical procedure, and the patient is regularly followed up for possible changes in the lesion.

**References**
