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Case report

Congenital infiltrating lipomatosis of the face with cerebral abnormalities

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Abstract. The aim of this study was to describe a possible variant of encephalo-craniocutaneous lipomatosis syndrome. Three cases of congenital infiltrating lipomatosis of the face, associated with cutaneous, subcutaneous, and cerebral abnormalities, are presented. This neurocutaneous syndrome appears very similar to encephalo-craniocutaneous lipomatosis syndrome but lacks the typical eye lesions.

Key words: Lipomatosis – Encephalocraniocutaneous lipomatosis – Neurocutaneous syndrome

Introduction

Congenital infiltrating lipomatosis (CIL) is a rare pathology that may cause a fatty mass infiltrating to the adjacent structures. Eighty percent of lipomatosis effects the lower limbs. Congenital infiltrating lipomatosis of the face is localized in the cheek, and physical examination reveals a poorly defined elastic tumor that may reach a remarkable size, with overlying skin of normal color and texture. Microscopic examination reveals non-capsulated fatty tissue formed by mature adipose tissue infiltrating the neighboring structures. The absence of lipoblastic proliferation, pleomorphism, and mitosis are important for the differential diagnosis of liposarcoma. Although they are benign, radical excision is difficult and the recurrence rate is very high [1]. Congenital infiltrating lipomatosis of the face associated with cutaneous, subcutaneous, and cerebral abnormalities are rare in the literature.

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Case reports

Case 1

A 2-year-old-male came to the hospital with a mass in the left cheek. The mass existed since birth and produced marked facial and mouth asymmetry. There were no relevant episodes during the pregnancy of his mother. Pigmented nevi were seen in the left cheek (Fig 1 a), left retro-auricular area, and left-anterior chest wall. In addition to the mass in the cheek, frontal and parietal subcutaneous masses were found at physical examination. These subcutaneous lesions were elastic and had ill-defined borders. Mental, neurological, and ophthalmological examinations revealed no abnormality. Magnetic resonance imaging revealed subcutaneous lipomatous infiltration of the left side of the face (Fig. 1 b). There were subcutaneous lipomas in the left frontal and parieto-occipital regions. The fatty tissue attacked the adjacent muscles and parotid gland. In addition, moderate hypertrophy of the left cerebral hemisphere, thickening of the left gray matter, enlargement of the left lateral ventricle, and two high-signal intensities in the white matter were seen (Fig. 1). Moderate unilateral megalencephaly was diagnosed.

The non-encapsulated adipose mass was excised as radically as possible. Invasion of the parotid gland was seen during the operation. The duct of parotid gland was damaged. Normal adipose tissue and invasion to the parotid gland were confirmed with histopathological examination (Fig. 1 d). After the operation, the face swelled and re-operation was conducted for repairing of the duct. The mass enlarged again 7 months later.

Case 2

A 3-month-old boy was admitted to the hospital with intractable seizures. The intrauterine life and his birth

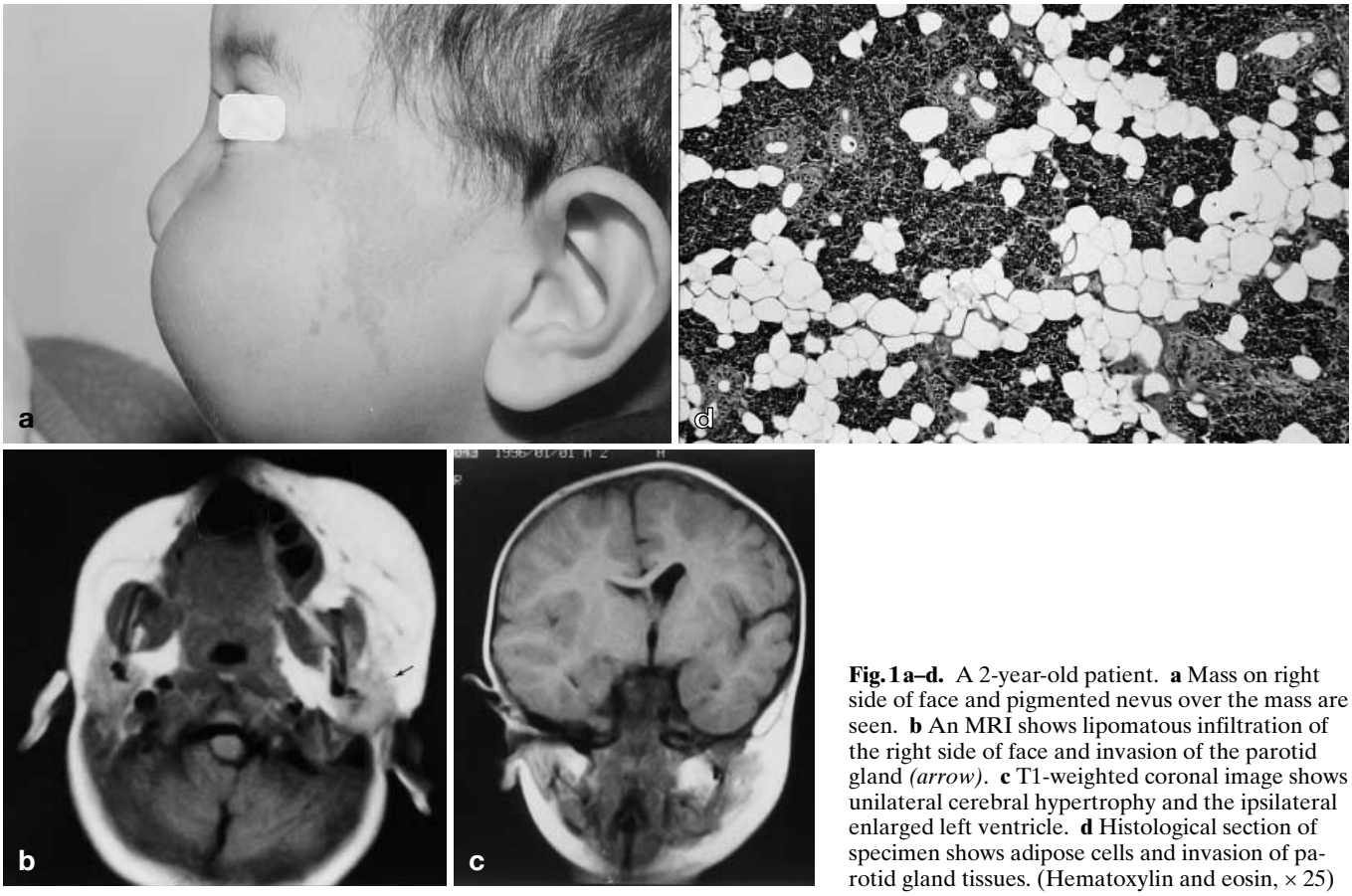


Fig. 1 a–d. A 2-year-old patient. **a** Mass on right side of face and pigmented nevus over the mass are seen. **b** An MRI shows lipomatous infiltration of the right side of face and invasion of the parotid gland *(arrow)*. **c** T1-weighted coronal image shows unilateral cerebral hypertrophy and the ipsilateral enlarged left ventricle. **d** Histological section of specimen shows adipose cells and invasion of parotid gland tissues. (Hematoxylin and eosin, × 25)

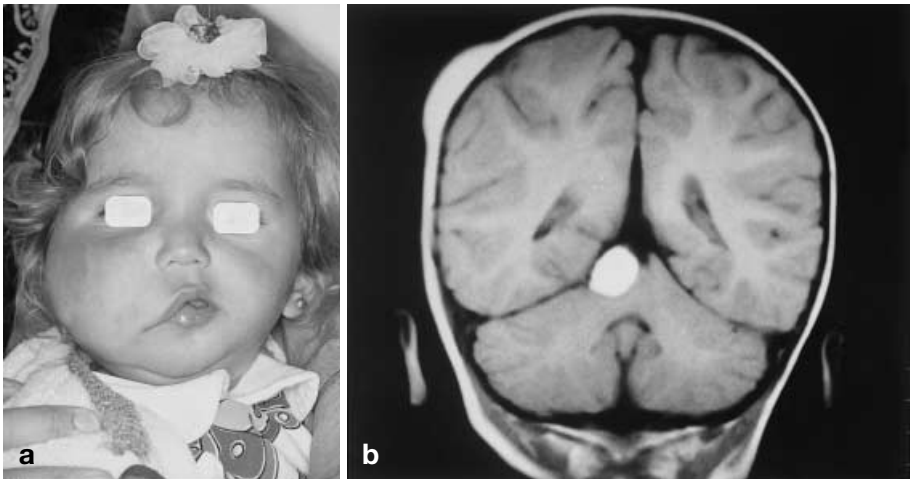
were normal. The mass on the left side of the face and cranial asymmetry were found at physical examination. Magnetic resonance imaging showed severe hypertrophy of the left cerebral hemisphere, unilateral diffuse pachygyria, enlargement of the left lateral ventricle, agenesis of corpus callosum, and high signal intensity on T2-weighted images in the white matter. Severe unilateral megalencephaly was diagnosed. Subcutaneous fatty mass in the left face infiltrated to the adjacent structures was shown on MRI examination. The patient died because of the status 4 months later.

Case 3

A 16-month-old girl presented with a subcutaneous mass in the right cheek since birth (Fig. 2a). There were no relevant episodes such as infectious disease, drug abuse, or exposure to teratogenic agent during the pregnancy. In addition to the elastic and ill-defined borders of mass in the cheek, an occipital subcutaneous mass was seen. There were no other physical abnormalities. In addition to subcutaneous, non-encapsulated adipose mass on the right side of the face, MRI showed occipital subcutaneous lipoma and extra-axial fatty mass in the quadrigeminal cistern (Fig. 2b). The fatty mass of the face was tried to be excised. The histology of the specimen was confirmed because normal adipose tissue was attacked in the parotid gland and the dermis. The intracranial extra-axial fatty mass was excised through a supracerebellar and infratentorial route. Lipoma was confirmed by histopathological examination. Mature adipose and heterotopic neural cells were seen in the pathological specimen (Fig. 2c). Six months later, the mass of the face grew again.

Discussion

Congenital infiltrating lipomatosis of the face is a rare but well-defined clinical entity [1]. Unilateral megalencephaly associated with CIL of the face is rare. Congenital infiltrating lipomatosis of the face and ipsilateral unilateral megalencephaly were seen in two of our cases. Unilateral megalencephaly is hamartomatous overgrowth of whole or part of the cerebral hemisphere. The disorder may also occur in patients with linear sebaceous nevus syndrome and neurofibromatosis type 1 [2]. Imaging studies show constants ranging from moderate to marked enlargement of the affected hemisphere. Abnormal T1 and T2 prolongation is seen within the white matter due to astrocytosis. The lateral ventricle on the affected side is always enlarged. Donati et al. [1] reported a case of CIL of the face with ipsilateral brain asymmetry (enlarged ventricle). There were intracellular inclusions of cytomegalovirus infection in the ipsilateral parotid gland tissue in his case. It did not exist in either of our cases.



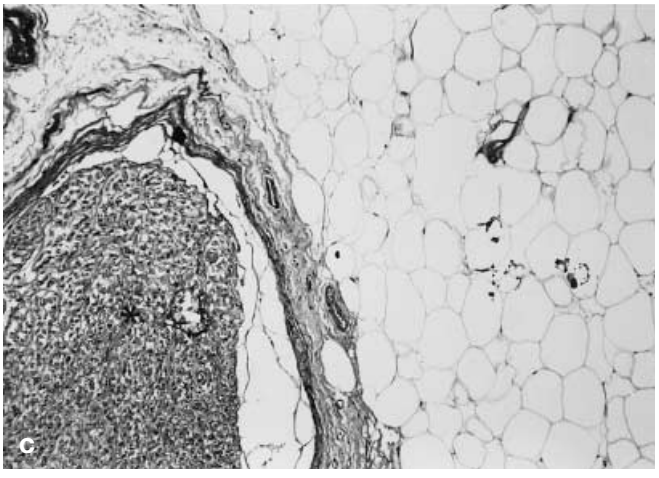


Fig. 2 a–c. A 16-month-old patient. **a** Mass on right side of the face is seen. **b** Intracranial and subcutaneous lipomas on coronal T1-weighted image. **c** Mature adipose and heterotopic neural cells *(asterisk)* are seen on histopathological examination of the intracranial lipoma. (Hematoxylin and eosin, × 25)

To our knowledge, CIL of the face with subcutaneous and intracranial lipomas has not been reported frequently in the literature [3]. Multiple subcutaneous lipoma were shown in two of our patients, and an intracranial lipoma was shown in one with CIL. Cranial lipomas are considered to be congenital malformations. Most of them are located in the midline region. The midline lipoma is clearly maldevelopmental, being frequently associated with structural anomalies of neighboring neural tissues [4, 5]. If the vault subsequently excluded the more embryologically related segment, multiple lipoma may be coexistent. Kodsi et al. [4] reported subcutaneous lipoma with slight enlargement of the left lateral ventricle. He had a temporal epibulbar choristoma of the left eye. Encephalocraniocutaneous lipomatosis (ECCL) syndrome was suggested in this case. Hennecam [5] reported scalp lipomas and cerebral malformations. He regarded this clinical picture as an overlap between ECCL and oculocerebrocutaneous syndrome.

Encephalocraniocutaneous lipomatosis is a congenital neurocutaneous disorder of the adipose tissue which is characterized by unilateral cerebral malformations and ipsilateral scalp, face, and eye lesions. The major manifestations include ipsilateral ocular, cutaneous, and central nervous system choristomas and hamartomas. Mental retardation, developmental delay, or seizures were present in most of the patients. The most common ocular lesions are epibulbar choristomas and connective tissue nevi of the eyelids. Alopecia was often found over or in close proximity to the lipoma; however, alopecia and ocular lesion were absent in the both cases. Central nervous system abnormalities was present in all patients. Porencephalic cyst, arachnoid cyst, lateral ventricle cyst, enlarged lateral ventricle, polymicrogyria, mass of the pontocerebellar angle, pachygyria, and intracranial lipoma may exist in such cases [4, 5, 6]. Ocular abnormalities were recorded in all patients. Fryer [7] reported a case with scalp lipoma, underlying bony skull defect and porencephaly. He also suggested that the clinical picture was compatible with ECCL, although there was no alopecia or scleral lesions. Although a spectrum of findings exist in ECCL, certain neurocutaneous findings appear in almost every patient. One of our patients had a hyperpigmented nevus near the eye. Another finding is that, in most cases in the literature, the neurocutaneous findings were ipsilateral. Single or multiple connective tissue nevi around the eyelids and "cafe-au-lait" spots (NF 1) with ECCL were reported [4, 8]. Three pigmented nevi were located unilaterally in our first case. We thought that ECCL may be associated with NF 1 in our first patient. Lesions in our cases were ipsilateral, except the midline intracranial lipoma in the third case.

Although there were no eye lesions, we thought that all of our cases were compatible with neurocutaneous syndrome, especially with ECCL syndrome. We think that it should be called a variant of ECCL. From a clinical point of view, patients who were admitted to hospital with a CIL of the face must also be checked for cranial pathology.

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Book review

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Hosten N., Liebig T.: Computertomographie von Kopf und Wirbelsäule. Stuttgart: Georg Thieme, 2000, 426 pages, 420 colour illustrations, 21 tables, DM 199.00, ISBN 3-13-117111-1

This book is part of a series entitled 'Reference Series: Diagnostic Radiology'. Although MRI has taken over many indications for sectional imaging of head and spine, CT is still considered a working procedure for numerous clinical indications. The authors have tried to provide a working book for the ideal use of CT of the head and spine, as well as considering its diagnostic advantages in comparison with MRI. The authors focus on the improved technologies for CT of the head and spine, three-dimensional reconstruction and the integration of CT information into computer-guided therapeutic modalities.

The book is clearly structured and provides tables and graphic representations of the most relevant imaging findings. Unfortunately, the images of normal anatomy are rather scarce and thus inexperienced readers may have difficulty in evaluating the material. The clinical case material is excellent, as is the picture quality. A more detailed description of the individual imaging findings would have helped young readers to gain a better understanding of some of the most important clinical questions.

All in all, the book is an important contribution to the literature concerning CT studies of the head and spine. I wish it success and well-deserved appreciation. T. Vogl, Frankfurt am Main

von Schulthess G. K.: Clinical positron emission tomography (PET): correlation with morphological cross-sectional imaging. Philadelphia: Lippincott Williams & Wilkins, 2000, 260 pages, illustrated, £ 87.00, ISBN 0-7817-1756-6

The strength of this textbook lies in the combination of morphological and functional imaging with special emphasis on PET imaging. The content on PET is competitive compared with other textbooks in the field but not superior. Overall, those who are looking for state-of-the-art knowledge will find it in a comprehensive format. However, this is not the first atlas of clinical PET to appear and its content does not contribute substantial new information.

The references are carefully selected and up to date. The PET images depicted in this atlas do not represent state-of-the-art images in oncology. For example, case examples for cancer imaging involve only non-attenuation-corrected PET images. Such images do not depict the true FDG distribution throughout the whole body or the part of the body imaged. In addition, state-of-the-art brain PET imaging involves normalization to the stereotaxic Tailarach space in order to match three-dimensional brain coordinates for comparison of morphology and function. Moreover, the quality of the cardiac images is variable.

For those who are looking for basic information, this book is valuable. If one is looking for more, it would not be a first choice.

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