Introduction

The Proteus syndrome is a rare hamartoneoplastic syndrome that may affect the brain, skull, and extracranial head and neck. We present a case with severe, characteristic findings. Brain abnormalities are not common in Proteus syndrome; when present, hemimegalencephaly and migrational disorders are typically seen, commonly with an associated seizure disorder. Maxillary and mandibular dysmorphism may occur, including unilateral condylar hyperplasia. Subcutaneous fatty, fibrous, lymphangiomatous masses commonly seen in this syndrome may involve the neck and face, leading to disfigurement and potential airway compromise.

Abstract The Proteus syndrome is a rare hamartoneoplastic syndrome that may affect the brain, skull, and extracranial head and neck. We present a case with severe, characteristic findings. Brain abnormalities are not common in Proteus syndrome; when present, hemimegalencephaly and migrational disorders are typically seen, commonly with an associated seizure disorder. Maxillary and mandibular dysmorphism may occur, including unilateral condylar hyperplasia. Subcutaneous fatty, fibrous, lymphangiomatous masses commonly seen in this syndrome may involve the neck and face, leading to disfigurement and potential airway compromise.

Key words Proteus syndrome · Magnetic resonance imaging

Case report

A girl first presented for investigation of an extensive facial mass at the age of 5 years. She had a history of a seizure disorder with “fair to good” control by a combination of phenobarbital, carbamazepine, and valproic acid. She had been found to have mental retardation and developmental delay. Examination demonstrated left hemifacial hypertrophy, macroglossia, and left facial palsy. She was sexually precocious, in Tanner stage II (pubic hair), at the age of 7 years.

MRI (Fig. 1) demonstrated a large, fibrofatty facial mass with infiltration throughout the left masticator, parotid, and parapharyngeal spaces. The left side of the tongue and masticator muscles were enlarged and diffusely infiltrated with fat, and contained numerous septa and flow voids. The genioglossus, geniohyoid, anterior belly of the digastric, and the mylohyoid muscles were involved. Left parapharyngeal infiltration encroached upon the airway. The left mandible, maxilla, zygoma, greater sphenoid wing, and frontal bone were hypertrophied, due primarily to expansion of the marrow space. The left mastoid air cells were poorly developed. Hyperostosis of the greater wing of the sphenoid distorted the orbital apex, and there was proptosis secondary to the sphenoid wing, maxillary, zygomatic, and frontal bone expansion.

The left temporal and occipital lobes were enlarged, along with the left hemicranium (Fig. 2). The left lateral ventricle was enlarged, the occipital horn being more affected than the temporal horn. The left parietal, temporal, and frontal lobe cortex and white matter were dysplastic, with loss of the normal gyral architecture. The left frontal lobe appeared pachygyric. The parietal and occipital lobes consisted essentially of a thin mantle surrounding the dilated lateral ventricle. The corticomedullary junction was indistinct, and the subcortical white matter had abnormal signal characteristics. Small cysts were seen in the dysplastic white matter. The left corona radiata and centrum semiovale appeared thin, as
Fig. 1a–c  Axial T1-weighted spin-echo images. a A large, infiltrating fatty mass is seen to extend into the tongue, enlarging its left half, but apparently respecting the deviated median raphe (white arrows). There is extensive septation, and several flow voids are seen within the mass. There is left mandibular hemihypertrophy (black arrow). b The fatty mass extends into the left parapharyngeal space (white arrow), encroaching upon the airway. There is extensive infiltration of the left masticator space and muscles of mastication (black arrow: infiltrated masseter). Marrow expansion of the maxilla results in its hemihypertrophy. There is also hypertrophy of the roots of the teeth, reflecting the different germ cell layers represented. Mandibular hypertrophy included the left condyle and ramus. c Hypertrophy of the greater wing of the sphenoid (black arrowhead), in conjunction with expansion of the zygoma (black arrow) and frontal bone (see Fig. 2), results in encroachment on the left orbit and proptosis.

Fig. 2a, b  Sagittal and axial T1-weighted spin echo. c T2-weighted fast spin-echo images. a The left lateral ventricle, including the temporal and occipital horns, is enlarged. The left frontal, parietal, and occipital cortex has abnormal gyration and signal characteristics. The left frontal bone is expanded (white arrow). b,c The left frontal lobe appears pachygyric (curved white arrow). The parietal and occipital lobes are markedly thin. The subcortical white matter gives abnormal signal, and small cysts are seen in the periventricular white matter.