A CASE OF PROGRESSIVE LIPODYSTROPHY
BARRAQUER-SIMON'S DISEASE

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INTRODUCTION

In its classical form (Mitchell, 1885) this rare disorder, found mainly in women, consists of a symmetrical and progressive loss of subcutaneous fat from the face and upper parts of the body only. The lower parts of the body are normal or may appear more obese than normal.

The aetiology of this disorder is obscure. Frank (1923) found the condition associated with osteogenesis imperfecta and regarded it as a defect of the whole mesenchyme. Langhof and Zabel (1960) concluded that the loss of fat is due to disturbed autonomic trophic regulations. Normal fatty tissue grafted by them to an area of lipodystrophy disappeared. Ellis (1962) has seen the condition develop rapidly in a little girl who had been terrified by a rat running over her face during sleep. An endocrine origin has also been suggested, and the condition variously attributed to dysfunction of the thyroid, pituitary or pineal. The only constant abnormality is almost complete absence of subcutaneous fat from affected areas. The dermis and epidermis appear normal; most patients show no other abnormality, but hyperthyroidism or an insulin-resistant diabetes is present in some. Harris and Reister (1940) observed a patient whose serum fat increased after a meal with a high fat content and who also had creatinurea and a lowered dextrose tolerance.

The disease is insidious in onset, beginning usually between the ages of 8 and 10 years. In a typical case it starts on the face, which gradually acquires an emaciated appearance with prominent zygomatic bones and hollow cheeks and temples. Subsequently, the upper extremities and upper trunk are affected, and the area of fat loss is sharply demarcated at the level of the iliac crest. Very rarely, the disorder starts at the iliac crest and gradually extends downwards to the legs.

No therapy exists for progressive lipodystrophy (Cecil and Loeb, Textbook of Medicine). The administration of various hormones had little effect, forced feeding resulted in excessive deposition of fat in unaffected areas. For cosmetic reasons injections of paraffin (Campbell, 1907) and human fat (Simons, 1911) have been made into the subcutaneous tissues of the face, but have caused only temporary improvement.

CASE REPORT

Miss R. F., a 19-year-old Art College student, was referred to Frenchay Hospital for improvement of her facial appearance. For the last eight years she had noticed she
was getting progressively thinner on the face and arms. Photographs of her showed that the disorder started after the age of 8 years (Fig. 1) and that it was well advanced by the age of 12 years.

The patient's general health was good with no history of any previous serious illness. Her brother and sister were normal in appearance and there was no relative known with the same condition.

On examination there was no other abnormality detected apart from the extreme emaciation affecting the face (Fig. 2) and upper parts of the body only.

**FIG. 1**
Patient at the age of 8 years. Normal facial contour.

**FIG. 2**
Facial appearance on admission. Prominent zygomatic bones and sunken cheeks.

**Details of Treatment and Operative Procedure.** Dental impressions were taken, in the usual way, and a silver cap-splint was made for the upper teeth. Silver trays were fixed to the splint by means of screws.

The splint was cemented in position the day before the operation which was carried out under general endotracheal anaesthesia and antibiotic cover. The mucoperiosteum in the upper buccal sulcus was incised down to bone from the upper right to upper left molar region across the middle of the dental roots. It was then reflected upwards and laterally to the anterior nasal spine, infraorbital margins and zygomatic buttresses, taking care to avoid damage to the infraorbital nerve. Two pieces of stent compound material were then warmed and moulded into the pouches so created. These, after cooling, were removed and clothed with a Thiersch skin graft which was obtained from the inner aspect of the thigh. Each piece of stent with the graft was inserted into the corresponding buccal pouch and held in position by the silver trays supporting them from below and the lip and cheeks falling over them. At the conclusion of the operation an Elastoplast strap was placed over the lip and cheeks to maintain the raw surfaces in close contact with the skin graft and minimise facial swelling during the succeeding two days.
The stent moulds were removed on the 18th day and were replaced by acrylic ones. The graft had taken satisfactorily and the buccal sulci seen to be adequately extended. The patient was discharged on the 24th day with instructions concerning the removal of silver trays and moulds for cleaning once a day.

The silver splints were removed two months later and the two acrylic moulds were replaced by a single prosthesis, which was found to be self-retentive.

The final result is shown by the photograph (Fig. 3) which shows the patient with the prosthesis in position.

![Facial appearance with prosthesis in position.](image)

**DISCUSSION**

One can well understand the psychological trauma associated with the extreme facial emaciation present in young females affected by this mysterious disorder. Our patient, in order to speed up her admission, wrote as follows: ‘... I feel that the delay in carrying out this promise (operation) is seriously affecting my general health and consequently the hope of success in my career’.

At present the only possible treatment is a cosmetic one, aiming in improving the facial appearance. This was achieved in the case reported by epithelial buccal inlays and the insertion of a self-retaining acrylic prosthesis. The procedure has the advantage of contouring the face at will by altering the size and shape of the acrylic prosthesis which can be incorporated into a partial or a full upper denture later in life.

The injection of human fat into the subcutaneous tissues of the face and the use of derma-fat grafts have resulted only in temporary improvement.

One of the silicone rubber materials such as silastic, now used in plastic surgery (Blocksma & Braley, 1965), could be used instead of fat, especially in building up hollow temples, and there may be a place for them in the future.
SUMMARY

A short account of the rare disorder progressive lipodystrophy is given and a classical case is presented. The patient's facial appearance was improved by extending and grafting the upper buccal sulcus into which a self-retaining acrylic prosthesis was inserted.

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