BPES was probably first reported by von Ammon in 1841 [1] and described more fully by Vignes in 1889 [2]. BPES is uncommon; the exact incidence is unknown. It is inherited as an autosomal dominant [3, 4] but 50% are sporadic.

Clinical features of BPES

The cardinal features, which primarily affect the soft tissues of the mid-face, are blepharophimosis, ptosis, epicanthus inversus and telecanthus.

The term ‘blepharophimosis’ refers to horizontal shortening of the palpebral aperture. It occurs in >100 syndromes. Since the orbital bones are normal in BPES [5] the interpupillary distance is also normal and the shortening occurs due to elongation of the medial canthal tendons and lateral displacement of the inner canthi. Mustarde proposed the term ‘telecanthus’ for this anomaly [5]. The lacrimal canaliculi and puncta are also abnormal [6].

The ptosis in BPES is always bilateral but may be asymmetrical and the levator function is usually poor [7]. Amblyopia occurs in >50% due to the ptosis or to coincident strabismus [7, 8].

Four patterns of epicanthic fold have been described [9, 10]. The first three, supraciliaris, palpebralis and tarsalis, arise from the upper lid and descend over the medial canthus to the lower lid. The fourth, inversus, arises from the lower lid and ascends into the upper lid and it is this type that characterises BPES. It is found in very few other syndromes. It is always bilateral.

Other periocular abnormalities in BPES include prominent brows, S-shaped upper lid, shortage of eyelid skin [9, 11-13] and hypoplasia of the plica and caruncle [14, 15].

Premature ovarian failure (POF) has been well recognised in BPES. Zlotogora [4] analysed this feature and proposed two types of BPES: Type I with POF and Type II with normal fertility.

Developmental delay, mental retardation and microcephaly have been described.
Surgical correction of BPES

The usual sequence of surgical treatment is correction of the epicanthic folds at about the age of 3-4 years and correction of the ptosis about 9-12 months later. Early surgery may be necessary for amblyopia [7, 8].

Epicanthic folds and telecanthus

Many operative techniques have been described for correction of epicanthic folds. Mustarde’s double Z-plasty [16] gives excellent results in most patients. At the same time subcutaneous fat in the inner canthal area must be reduced and the medial canthal tendon must be shortened [17, 18]. The aim is to reduce the intercanthal distance to about half the interpupillary distance [19].

Ptosis

The ptosis is usually corrected with a Crawford brow suspension procedure with autogenous fascia lata [18]. However, a non-autogenous material such as a silicone rod is preferred for young children having early surgery for amblyopia.

Other surgery

Other surgery is occasionally required. If the eyelid skin is tight a full thickness skin graft may be required.

Results of surgery

In an analysis of the improvement with surgery Taylor [19] found that Mustarde’s double Z-plasty completely abolished the epicanthic folds, there was a 26% reduction in intercanthal distance and all had upper lid folds. Medial canthal scarring was visible but mild in 50%.

Genetics of BPES

The genetics of BPES have been investigated extensively. Vignes in 1889 [2] recognised that BPES ran in families and Dimitry in 1921 [2] observed that it was dominantly inherited. In 1976 Moraine [20] first noted the link between BPES and female infertility. Suddenly the syndrome became far more significant and a search for a gene began. By 1995 the locus had been identified as 3q23 [21-23] and the gene was finally identified by Crisponi and co-workers in 2001 [24]. BPES is due to mutations within, or influencing, a single
gene, the FOXL2 gene. Well over 100 intragenic mutations have been described. Many attempts have been made to link the mutations with variations in the phenotype but with only partial success [25]. Deletions involving the gene result in more widespread and severe anomalies [26, 27].

**Risk of POF**

Assessment of the risk or POF can be helped by the family history. Molecular genetic testing may be helpful in some families. Females with BPES should all see a clinical geneticist and, during puberty, an endocrinologist to assess the onset of POF.


