BPES : An Overview

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- The blepharophimosis ptosis epicanthus inversus syndrome (BPES, also known as Waardenburg syndrome) was probably first reported by Ammon in 1841 and described more fully by Vignes in 1889. Its primary effects on the soft tissue of the midface are blepharophimosis, ptosis, epicanthus inversus and telecanthus.
- It is a genetic disorder usually inherited in an autosomal dominant manner whose main features are the abnormal shape, position and alignment of the eyelids. Malformation of the eyelids may severely impair visual function. BPES is sometimes associated with developmental delay, but patients with BPES typically have a normal lifespan.

Its **diagnosis** is based on four major features present at birth:

1. Short horizontal palpebral fissures (Blepharophimosis)
2. Drooping of the eyelids (Ptosis)
3. Vertical fold of skin from the lower eyelid up either side of the nose (Epicanthus Inversus)
4. Lateral displacement of the inner canthi with normal interpupillary distance(Telecanthus)

Two types of BPES are recognized:

> Type I BPES includes the four major eyelid features and female infertility as a result of premature ovarian failure.

> Type II BPES consists only of eyelid abnormalities.

Laboratory test

- Genomic DNA extracted from peripheral blood.
- PCR direct sequencing and quantitative real-time PCR-based copy number screening for the whole exon of FOXL2 has to be performed.
- Deletions and mutation in the FOXL2 gene in BPES patients detected by q-real-time PCR technique which enriched the diagnostic methods of molecular genetics.

Differential diagnosis

1. Congenital ptosis.
2. Isolated blepharophimosis.
3. Other dysmorphic syndromes.

Management

- Management of BPES is primarily surgical if indicated. Care should be given to treat assoicated amblyopia.
- 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.
- EPICANTHUS FOLD AND TELECANTHUS : double Z or Y-Z plasties, Transnasal wiring of the medial canthal tendons. One stage correction of the telecanthus, epicanthic folds and the ptosis has been reported.
- PTOSIS: Usually is corrected with a brow suspension procedure.
- PRIMARY OVARIAN FALURE: Various pharmacological therapies have been shown to be ineffective .

  Embryo cryopreservation.

  Hormone replacement therapy to reduce the early post-menopause effect.

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