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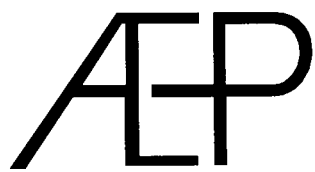
GUNNAR LENNERSTRAND

Department of Ophthalmology
Karolinska Institutet
Huddinge University Hospital
Stockholm, Sweden



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CONGENITAL INNERVATION DEFECT SYNDROME

Ahmed A. Assaf, MD, FRCOphth
Department of Ophthalmology
Stoke Mandeville Hospital, Aylesbury, Bucks, UK

ABSTRACT

Recent evidence suggested that normal intra-uterine development of the innervation of the extra-ocular muscles (EOMs) is of critical importance for normal development of these muscles themselves. Congenital defects in the innervation of EOMs can result in abnormal muscle structure depending on the stage and the extent of such innervational defects.

The current accepted view of Duane's retraction syndrome (DRS) and monocular elevation deficiency (MED) or double elevator palsy fits into such category. Congenital loss of normal innervation of the lateral rectus muscle, as seen in DRS can result in abnormal muscle structure, loss of muscle function, co-contraction, and contraction against a tight muscle. A more recent view is that congenital fibrosis syndrome (CFS) belongs to this category of congenital innervation defect (CID). In this instance involving the III cranial nerve and it's supra-nuclear complex. These affect the vertical, and to a lesser degree the horizontal, eye movements.

These conditions will be linked in a proposed classification under the title CID syndrome, with subclasses, for easier identification of the spectrum of this syndrome. Such proposition will be presented and discussed.

Key Words: Congenital innervation defect syndrome, Duane's retraction syndrome, congenital fibrosis syndrome of the extra-ocular muscles, double elevator palsy (monocular elevation deficiency).

CONGENITAL INNERVATION DEFECTS

Recent evidence suggests that normal intra-uterine development of extra-ocular muscles is dependent upon normal development of innervation.[1] Thus congenital loss of innervation can result in muscle structural abnormalities, the likes of which are seen in DRS.

Additionally, chronic loss of innervation can produce histological effects similar to that seen in primary muscle myopathy.[2]

For such congenital ocular motor defects associated with congenital loss/abnormality of innervation I would like to propose the term of *Congenital Innervation Defect (CID) syndrome.* The features of this syndrome are:

- Congenital Defect in the innervation EOMs.
- Present since birth, and non-progressive.
- Unilateral or bilateral.
- Findings are not explained by pure isolated oculo-motor nerve palsy/palsies.
- Anatomical muscle changes, including tight muscle.
- Can be associated with synkinesis phenomena and/or co-contraction.
- Abnormal head posture is common.

Such congenital neurogenic disorders of extra-ocular muscles are not uncommon. This syndrome includes DRS and MED. More recently, congenital fibrosis syndrome is increasingly considered as neurogenic rather than muscular in aetiology and can be considered among this category.

It is universally accepted now that Duane's retraction syndrome is the result of abnormal innervation of the Lateral rectus muscle. This can result from either hypoplasia of the VI nerve nucleus or miswiring in the peripheral innervation itself arising from the III cranial nerve.[3] The evidence for neurogenic aetiology was first derived from co-contraction elicited by electrophysiological studies.[4,5] Additionally, the association of DRS with synkinesis phenomena such as Marcus Gunn jaw-winking have added further evidence.[6] Further evidence was derived from pathological studies.[7,8] Recent advances in the imaging technology have made it easier to detect such subtle anatomical changes.[9]

Monocular elevation deficiency (double elevator palsy) has also been recognised to be associated with disorders of innervation beside that of anatomical abnormalities. This has been observed in acquired paralysis resulting in MED.[10,11] Significant number of patients with congenital MED is associated with synkinesis phenomena such Marcus Gunn jaw-winking.[12,13] Additionally, Olson and Scott[13] found DVD was present in 29% of MED patients perhaps suggesting innervational rather than anatomical aetiology.

Congenital fibrosis syndrome of the extra-ocular muscles is becoming more recognised as being caused by an innervational rather than a primary

defect of the extra-ocular muscles. Like Duane's syndrome, CFS is characterised by abnormal eye movements, tight extra-ocular muscles, and muscle co-contracture. On this occasion, it is suggested that there is involvement of the vertical eye movements which may have a horizontal element incorporated. The innervational aetiology here is derived from many aspects: anatomical, histological and clinical.[14] More recently, Engle etal[15] has demonstrated an absent superior division of III cranial nerve in a patient with presumed CFS. Beside this CFS is also reported to be associated with synergestic divergence and Marcus Gunn jaw-winking.[16] EMG studies in Möbius syndrome, which is a sub-type of CFS, have shown patterns suggesting of supra-nuclear origin.[17,18]

CLASSIFICATION

Based on the evidence discussed above I have suggested a classification for this CID syndrome.[19] This classification would incorporate the present entities of Duane's retraction syndrome, double elevator palsy, and CFS. Such classification would include the following sub-types:

Type I CID: only horizontal eye movements are involved. Subtypes: *A*, *B*, and *C*; A = loss of Adduction, B = loss of aBduction, and C for both. This type is the current DRS.

Type II CID: only vertical eye movements are involved. Subtypes: *A* = upgaze (commonest), *B* = downgaze, and *C* = both upgaze and downgaze.

Type III CID is a combination of vertical and horizontal motility disorder.

Subtypes: *A1, A2, A3* = upgaze combined with horizontal involvement.
Limited adduction = 1, limited abduction = 2, both = 3.

Subtypes: *B1, B2, B3* = downgaze combined with horizontal involvement (1, 2, and 3 as above).

Subtypes: *C1, C2, C3* = upgaze and downgaze combined horizontal involvement (1, 2, and 3, as above).

All types can be unilateral or bilateral.

This terminology and classification would first encompass such diverse entities into their joint aetiology. Secondly, it will make it much easier to understand, remember and plan therapy of such complex ocular motility disorders.

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