A RARE CASE OF BILATERAL DUANE RETRACTION SYNDROME WITH BILATERAL CEREBELLAR ATROPHY

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Abstract:
Duane's retraction syndrome is characterized by globe retraction and palpebral fissure narrowing on adduction, with restriction of abduction or adduction or both. The proposed mechanism for the impaired ocular motility is congenital miswiring of the lateral and medial rectus muscles. Electromyographic studies show decreased or absent firing of the lateral rectus and medial rectus during attempted adduction causing globe retraction. It may be associated with other ocular as well as systemic congenital anomalies such as sensorineural deafness, upper limbs defect, facioauriculo-vertebral anomalies, cardiac and genitourinary anomalies. We report a case of rare association of bilateral Duane retraction syndrome with bilateral cerebellar atrophy in a 28 year-old male. History, the ocular motility examination and neurological examination revealed that our patient has congenital bilateral Duane retraction syndrome with bilateral cerebellar signs. We concluded that there is novel association between Duane retraction syndrome with chronic progressive bilateral cerebellar degeneration.

Keyword: Duane retraction syndrome, cerebellar atrophy, abduction, adduction

HISTORY
A 28 years old male came to outpatient department with history of slurring of speech - 8 years Difficulty in walking, clumsiness of hands, head nodding - 4 years

History of present illness
Eight years back patient was apparently normal then he has difficulty in speech which is insidious in onset gradually progressive now static for 3-4 years. History of irrelevant behaviour, irrelevant talk present 4 years back for which patient took treatment in psychiatric OP now it is decreased. Difficulty in walking for 4 years which is onset and slowly progressive associated with wide based gait and head nodding. After 6 months of onset he has clumsiness of hands in the form of difficulty in holding objects, spillage of food while eating, intentional tremor, and tremulousness of the body.
No history of swaying to either side, no history of difficulty in negotiating in narrow pathways. No history of weakness of limbs, no history of suggestive of sensory abnormalities. No history of other cranial nerve symptoms except for dysarthria. No history of defective vision, or diplopia.

No history suggestive of autonomic disturbances.

**Past history**

History of abnormal eye position present since childhood. No history of diabetes mellitus, hypertension, cardiac problems, or epileptic. No history of previous similar illness.

**Personal history**

Not a smoker or alcoholic. Taking mixed diet.

**Family history**

He is born out of non-consanguineous marriage. He has two younger brothers. They are normal no neurological illness.

**PHYSICAL EXAMINATION**

He is conscious, oriented to time place and person, moderately built and nourished. Not anaemic, not jaundice, no pedal oedema. Jugular venous pressure is normal. No skeletal deformities and no neurocutaneous markers. Vitals – stable.

**Higher mental functions examination:**

*MMSE score – 26*

Speech – staccato speech.

**Cranial nerve examination:**

*Olfactory nerve – intact*

Optic nerve – visual acuity - 6/12, near vision - normal. Colour vision – normal. Fundus normal.

Oculomotor, trochlear and abducent nerves:

*Extraocular movements:*

Narrow palpebral fissure and no strabismus in primary gaze.

During right horizontal gaze, further narrowing of palpebral fissure (enophthalmos). During left horizontal gaze, in the right eye, adduction is present and further narrowing of palpebral fissure (enophthalmos) but in the left eye there is no abduction and no change in palpebral fissure.
Vertical gaze - normal on both sides. Pupil-3mm size equal on both sides. Direct Light reflex and consensual light reflex present bilaterally. Trigeminal nerve, facial nerve, vestibulocochlear nerve, glossopharyngeal nerve, vagus nerve, accessory nerve and hypoglossal nerve are intact.

**Cerebellar system examination:**
- No nystagmus, staccato speech- present, titubation- present, finger nose and finger nose test- positive on both sides. Dsdiadocokinesia present on both sides. Dysmetria and intention tremors present on both sides. Heel shin test- positive. Ataxic gait- present. **Spinomotor system examination:**
  - Bulk- normal, tone- hypotonia, power- 5/5, all deep tendon reflexes normally present on both sides. Plantar reflex - Bilateral flexor response. Superficial reflexes present.
  - All modalities of sensations are intact. Rombergs sign-negative

**Discussion:**
- Congenital limitation of abduction and/or adduction
- Globe retraction (co-contraction) on adduction
- Palpebral fissure (i.e., the separation between the upper and lower eyelids)

Most individuals with Duane syndrome have isolated Duane syndrome, i.e., they do not have other detected congenital anomalies. The vast majority of individuals with isolated Duane syndrome are simplex cases (i.e., single occurrence in a family) Duane syndrome can be clinically subdivided into three types (Huber classification):

Type 1 (~75%-80% of all Duane syndrome) is characterized by the following:
- Absent to markedly restricted abduction
- Normal to mildly restricted adduction
- Retraction of the globe and narrowing of the palpebral fissure on adduction
- Upshoot and downshoot of affected globe on attempted adduction
- Esotropia in primary gaze (variably present)
- Head turn toward involved side (variably present)

Type 2 (~5%-10% of all Duane syndrome) is characterized by the following:

- Unilateral or bilateral involvement
i. Absent to markedly restricted adduction
ii. Normal to mildly restricted abduction
iii. Retraction of the globe and narrowing of the palpebral fissure (the separation between the upper and lower eyelids) on adduction
iv. Upshoot and downshoot of affected globe on attempted adduction (variably present)
v. Esotropia in primary gaze (variably present)
vi. Head turn toward uninvolved side (variably present)
vii. Unilateral or bilateral involvement

Type 3 (~10%-20% of all Duane syndrome) is characterized by the following:
i. Absent to markedly restricted abduction
ii. Absent to markedly restricted adduction
iii. Retraction of the globe and narrowing of the palpebral fissure on attempted adduction
iv. Upshoot and downshoot of affected globe on attempted adduction (more common than in types 1 or 2)
v. Esotropia or exotropia in primary gaze (variably present)
vi. Head turn toward involved side (variably present)
vii. Unilateral or bilateral involvement

Pathophysiology
Duane syndrome is a strabismus syndrome characterized by congenital non-progressive horizontal ophthalmoplegia primarily affecting the abducens nucleus and nerve and its innervated extraocular muscle, the lateral rectus muscle. At birth, affected individuals have restricted ability to move the affected eye(s) outward (abduction) and/or inward (adduction). In addition, the globe retracts into the orbit with attempted adduction, accompanied by narrowing of the palpebral fissure. Duane syndrome results from maldevelopment of motor neurons in the abducens nucleus and aberrant innervation of the lateral rectus muscle. Electromyography revealed simultaneous activation of the medial and lateral rectus muscles, supporting co-contraction of these two horizontal muscles as the cause of the globe retraction. Magnetic resonance imaging (MRI) in simplex cases has the absence of cranial nerve VI.

Duane syndrome can be associated with restriction in vertical movement of the eyes, strabismus, Amblyopia, Marcus Gunn jaw-winking phenomenon. Visual acuity is good except in those individuals with amblyopia.

It can also be associated with congenital anomalies like Okihiro syndrome, Duane-radial ray syndrome, acro-renal-ocular syndrome, (SALL4 gene mutations). These overlapping syndromes are characterized by unilateral or bilateral Duane syndrome and radial ray malformations that can include thenar hypoplasia and/or hypoplasia or aplasia of the thumbs; hypoplasia or aplasia of the radii; shortening and radial deviation of the forearms; triphalangeal thumbs; and duplication of the thumb (preaxial polydactyly). Deafness, renal anomalies, and imperforate anus can be co-inherited. Inheritance is autosomal dominant.
Townes-Brocks syndrome results from mutations in SALL1 characterized by renal, anal, limb and ear anomalies and is an autosomal dominantly inherited malformation syndrome.

Bosley-Salih-Alorainy syndrome, Athabascan brain stem dysgenesis syndrome (HOXA1 mutation) characterized by Duane syndrome type 3 or horizontal gaze palsy and, in most individuals, bilateral sensorineural hearing loss caused by an absent cochlea and rudimentary inner-ear development.


Our patient belongs to type-1 Duane retraction syndrome (bilateral) and associated with bilateral cerebellar atrophy. He has no other congenital anomalies. **Conclusion:** There is novel association between bilateral Duane retraction syndrome (type-1) with bilateral cerebellar degeneration.

**References:**


