Case Report

A case of ataxia

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- **58 year male precnt with complain of insidious onset of slowly progressive, asymmetric tremulousness of both hands during activities in last 8 years. He also complain of imbalance in gait for last 3 years with tendency of fall for last 2 months.**
- **In last 1 year changes of speeches with separation of syllables.**

History of Present illness :

A 58-year-old right-handed gentleman from Asansol, nondiabetic, nonhypertensive and an accountant by profession came to our OPD with complaints of insidious onset slowly progressive tremulousness of both hands present mainly during activities for last 8 years. It had asymmetric onset in the right hand, involving the left hand after 6months and head was affected after another one year. It was present mainly during writing, holding some objects or in out stretched arms but not at rest or sleep. In last two years he has developed exacerbation of tremor intensity on reaching some target leading to hand-mouth incordination. He also complained of imbalance in gait with tendency to sway towards either side while walking since last three years along with three episodes of fall in last two months. Clumsiness of gait was present similarly both during day and night. There was no side predilection, no loss of consciousness, vertigo or lightheadedness associated with those falls and every time he tried to protect himself with hands. He had no difficulty in turning in bed and could raise his limbs and neck from bed while lying on it. At present, though his daily activity is significantly affected by tremor and loss of balance, he is still able to walk without support. In last one year there is a change in her speech in the form of undue separation of the syllables with inappropriate pressure on some of them. He does not have numbness or tingling, any sensory loss, no bowel or bladder involvement, no history suggestive of any involvement of cranial nerves or cognitive decline, loss of consciousness or seizures. He is unmarried, on Indian vegetarian diet, has normal sleep and appetite. He smokes 5-6 cigarettes per day and is a social drinker. No major improvement of tremor was seen after drinking. His family history revealed similar tremor and imbalance in his father and one of his paternal uncles had psychiatric illness. He is on propranolol and trihexyphenidyl for last three years without much improvement. He has no history of other major systemic illness or significant weight loss.

Examination :

The patient was of normal built and nutrition. His heart rate was regular at 86/min, Blood pressue 116/76mm of Hg without any postural drop. He was well oriented with 28/30 in MMSE scale. Speech was fluent with scanning character. Extraocular movements were full but there was gaze-evoked nystagmus. Saccades were slow and persuit broken. The rest of the cranial nerve examination was normal. There was no atrophy. Activated cogwheel rigidity in both upper limbs and spasticity in both lower limbs was noted. Action, postural and Intention tremor in both upper limbs and titubation were seen. Strength is 5/5 in all four limbs. Generalized hyperreflexia was found. The plantar response was extensor bilaterally with absent abdominal and cremesteric reflexes. Sensation was intact to light touch, pinprick, temperature, vibration and joint position sense. There is ataxia bilaterally more on finger-to-nose testing than on heel-to-shin testing with dysdiadochokinesia. He had a broad based ataxic gait with good stride length but arm swing was mildly reduced bilaterally. He could not perform tandem gait.

Investigation :

Routine laboratory tests including FBS, lipid profile, ceruloplasmin, HIV1&2, VDRL, thyroid studies normal. Anti GAD Ab, anti Hu and anti TTG was negative. Nerve Conduction Study was normal. KF ring was absent.MRI of the Brain shows marked diffuse cerebral and cerebellar atrophy with normal cervical cord screening. Genetic testing for SCA panel revealed pathologic CAG triplet repeat in PPP2R2B gene confirming the diagnosis of SCA 12.

Discussion :

Illness in this given case started with postural and kinetic tremor of hands without any rest tremor. Later on there was history of head tremor (titubation) and intention tremor (tremor which increased in the terminal part of a goal directed activity) along with history of dysmetria (past pointing and hand mouth inco-ordination). Anatomical site for postural and action tremor is Gullian-Mollaret triangle in brainstem and cerebellar outflow tract. Intention tremor is anatomically localized to the ipsilateral dentate nucleus and rubro-olivo-cerebellar circuit. For incordination and dysmetria ipsilateral cerebellar hemisphere is involved[1].

Next complain he made was of imbalance in stance and gait along with clumsiness of movements. In absence of any definite history of overt weakness (normal movement of limb and trunk in lying down position) the phenomenology is probably *ataxia.* This can be caused by either by cerebellar dysfunction or proprioceptive dysfunction. In our case, as ataxia is present both during day and night without any history of undue exacerbation in night, cerebellar ataxia due to involvement of rostral vermis is more likely.

There is history three episodes of fall with preserved postural reflex and without any LOC, presyncopal or vestibular symptom.

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This is more likely to be due to imbalance although mild limb weakness can't be ruled out.

Speech abnormality like irregular breakdown of speech with explosions of syllables interrupted by pauses, known as scanning speech is due involvement of the left superior paravermian area[1].

Hence by summarizing the historical analysis we can comment that there is suggestion of involvement of Gullian-Mollaret triangle in brainstem and cerebellum both at midline and bilateral hemisphere or its connection.

After examination we have confirmed features of affection of Gullian-Mollaret triangle in brainstem and pancerebellar involvement (gait and stance ataxia for midline rostral vermis, scanning speech for paravermian area, appendicular ataxia, tremor for cerebellar hemisphere, gaze evoked nystagmus for vestibulocerebellum) with suggestion of additional anatomical sites involvement in the form of pathways of saccadic and pursuit eye movement, bilateral corticospinal tract (spasticity in lower limbs, hyperreflexia and abnormal superficial reflexes), bilateral basal ganglia and its connection(cogwheel rigidity, decreased arm swing). So this is a case of chronic cerebellar ataxia with additional features of other neuraxis involvement.

Similar to every other case in neurology clues to etio-pathogenesis of the ataxic disease also lie in analysis of duration and course of illness, associated features, family history, dietary, drug and toxin history. In this case insidious onset, slowly progressive course and long duration make neurodegenaration as the most likely pathology. Among this group autosomal dominant cerebellar atxia (Spinocerebellar ataxia-SCA), late onset autosomal recessive cerebellar ataxia(odd point is positive family history in preceeding generation), Parkinson plus syndrome like multisystem atrophy-cerebellar type(MSA-C), Wilson disease(no KF ring was seen); Fragile-X associated tremor ataxia syndrome(FXTAS) are possible etiologies. Among other pathogenesis chronic drug (there is no h/o chronic medication use) and toxin exposure like Alcoholic degeneration (amount of intake here is nominal to cause cerebellar degeneration); paraneoplastic cerebellar degeneration like in Hodgkins lymphoma(no systemic symptoms and long history), immune mediated codition like gluten ataxia(no h/o dirrhoea and gluten intolerence), anti GAD Ab mediated atxia(usually have subacute course and most common in female: not matching with this case), chronic infection like Syphilis, HIV(no systemic symptoms.); very slow growing cerebellar tumour or multiple granuloma, vascular pathology like multi-infarct state, vasculitis (ruled out by neuroimaging) are among rare possibilities. Other pathologies like stroke, demyelination and intoxication have acute presentation and can't be considered here. If we take into account other associated features then conditions that can present with chronic cerebellar ataxia with tremor are spinocerebellar ataxia type 2,3, 12, 15, FXTAS, MSA-C or it may be a case of cerebellar atxia with essential tremor(ET). But asymmetric onset, poor response to alcohol makes ET unlikely. Among others; MSA-C usually have prominent autonomic features, FXTAS is usually have a much late onset in eighth decade, SCA 2 have very slow saccades and hyporeflexia. Family history of psychiatric illness and cognitive decline favors diagnosis of SCA 12[2,3]. This was proved by genetic test report showing CAG repeat in the PPP2R2B gene. After review of literature it was seen that this disease is particularly common in Indian subcontinent in the Agarwal community because of inbreeding. This gentleman in our case was also from that same community.

Conclusion

The idea of presenting this case is to emphasize few atypical features of SCA 12 like its presentation with postural or action tremor, relatively milder form of atxia, less prominence of anticipation in family history, presence of psychiatric illness and cognitive decline in the patient or in family members and last but not the least its its common occurance in Agarwal community of Indian subcontinent[4,5].

References

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Editorial Comments :

- **Ataxia, a Greek word meaning "lack of order".**
- **Disorders affecting the cerebellum, its connections, and the afferent proprioceptive pathways.**
- **Inability to perform a motor activity.**

Key words : **Ataxia, cerebellum, mortor activity.**

Ataxia, a Greek word meaning "lack of order", is the predominant manifestation of many acquired and inherited neurologic disorders affecting the cerebellum, its connections, and the afferent proprioceptive pathways. History of ataxia is usually very straight forward but at times the patient may complain of inability to perform a motor activity and relate it to limb weakness which the clinician can't find. So in a patient who can identify a tool, plan its use properly, has no motor weakness, ataxia, sensory or cerebellar, should always be considered as the cause of his/her inability to carry out the activity. We will discuss the clinical approach to an ataxic patient with reference to the case presented below.