

Ekboms Syndrome (Restless Legs Syndrome): 2 Case Report And Literature Review

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ABSTRACT:

BACKGROUND: Ekboms syndrome is an uncommon, chronic disorder that often has a familial basis, with evidence of autosomal dominant inheritance. It occurs in about 5% of adults and about one third of patients would have multiple affected family members.

AIM AND OBJECTIVE: This report is intended to increase the index of suspicion of clinicians to this disease for prompt diagnosis and avoidance of unnecessary expensive neurological investigation.

CASE SUMMARY

This case report is on 2 (two) adult males (Mr. T.A and Mr. F.A) who were 43 and 38 years respectively and presented at the clinic in Jahun General Hospital with irresistible movement of the lower limbs at rest. This started a year prior to presentation when they both began feeling needle sensation on both legs starting from the feet (paraesthesia/dysaesthesia) until they noticed their legs start to move while at rest but relieved on moving it or walking.

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INTRODUCTION

Ekboms syndrome (restless legs syndrome) is an autosomal dominant disorder and has a familial basis in about one third of patients.

The pathogenesis is said to be a defect in the basal ganglia following an endogenous opioid system fault which causes altered central pain processing. Research and brain autopsies have implicated both dopaminergic system and Iron insufficiency in the substantia nigra. Iron is an essential cofactor for the formation of L-dopa, the precursor of dopamine. Six genetic loci found by linkage have been implicated which are-chromosome 12q, 14q, 9p, 20p, 2p and 16p12.1 respectively. Three genes, MEIS1, BTBD9 & MAP2K5 were found to be associated to restless legs syndrome though their role is still unclear.

It is a sensori-motor disorder characterized by an unpleasant creeping (shooting or tingling) sensation that arise deep within the legs and starting from the feet and occasionally in the arms especially when the patient is relaxed. It is said to occur in 5% of adults with a female preponderance of about F:M (2:1). Periodic leg movements can also occur during sleep and can be documented by polysomnography. The paraesthetic feeling stops during movement of the limbs in contrast to peripheral neuropathy of other causes.

The paraesthesia is worsened by pregnancy, caffeine and sleep deprivation. There are two types which are primary and secondary:-

Primary restless legs syndrome: Is that which occurs without any attributable cause seen.

Secondary restless legs syndrome: Is that caused by other disease or substances such as renal failure, pregnancy, rheumatoid arthritis, diabetes mellitus, iron deficiency anaemia, chronic Obstructive Pulmonary Disease (COPD), malignancy, varicose veins, Celiac disease, Sjogrens syndrome, Thyroid disease, Parkinsons disease, Chronic liver disease, Sleep apnoea syndrome, Fibromyalgia and Magnesium deficiency. Certain medications may cause or worsen restless legs syndrome: - antiemetics mostly the antidopaminergic (prochlorperazine, metoclopramide), antipsychotics, anticonvulsants, antihistamines, antidepressants (older tricyclic antidepressants and newer selective serotonin receptor inhibitors) and rebound effect of benzodiazepine withdrawal (opiate withdrawal may also "trigger" the syndrome).

The syndrome is sensitive to dopaminergic drugs such as L-dopa, and dopamine agonists. Benzodiazepines and certain anticonvulsants such as diazepam, clonazepam, narcotics such as codeine, oxycodone, propoxyphene can also be used.

CASE REPORT

CASE 1

A 43 year old male who came to the hospital because he started feeling embarrassed that his legs were moving while he was seated with his friends and sitting relaxed in his home. There was no history of cough, drooling of fluid from nose, mouth or night sweats or loss of weight. There was no history of yellowness of eyes, vomiting and swelling of the limbs. No history of trauma, polydipsia, polyphagia and polyuria or whitish lesion or excoriation/cicatrisation marks on the body. There was occasional intake of cassava. He was not a food choosing person but ate anything that was served. Not a strict vegetarian, not an alcoholic or smoker. No history of incontinence of urine or faeces, phobia, anxiety disorder, headache or sweating palms in him. He was married with two (2) wives with eight (8) children between them and the fifth (5) in a family of nine (9) children. The father is late from an unascertainable cause but no history of restless legs prior to his death. All siblings, mother and his own children are all alive with no such complaint. He was a farmer by occupation.

Examination on him revealed a middle -age man, well

kept, anxious looking, not pale, afebrile, anicteric, no lymphadenopathy, no pigmentation or whitish lesion (uraemic frost) on the body, no excoriation, no finger clubbing, no oedema but movement of legs at rest which stopped on moving about. The restless legs could not be stopped voluntarily. Fine and deep touch were preserved. Position and vibration sense were also preserved. The chest was clear and no murmur was heard. There was no gibbus on palpation of vertebral column and no musculoskeletal system abnormality. He was oriented in time, place and person. Insight, judgement, memory (Long and short term), attention, arithmetic and abstract thinking were all intact.

RBS = 8 mmol/l, and BP = 110/70 mmHg & PR = 70 beats/min. PCV = 38%, FBC microscopy and the differential was normal, RVD - negative, S/E/U/CR normal, LFT - was normal, Cxray (PA) - was normal. X ray of lumbosacral (AP and lateral) was normal.

A tentative diagnosis of Ekboms syndrome was made. Patient was placed on Amitryptiline of 75mg nocte for a duration of one year. I followed him up initially at an interval of two weeks' follow up visit with mild improvement, was latter placed on two (2) months' follow up visit and subsequently placed on four (4) months' follow up visit. At the last visit a year after presentation; the patients had marked improvement with mild movement of the legs at rest and occasional paraesthesia.

CASE 2

A 38 year old male who presented with irresistible movements of the legs while at rest, chest pain of two (2) months' duration which was dull, griping and intermittent in nature and headache of one (1) months' duration which was frontal in nature precipitated by waking from sleep, daily activity, exposure to radiation from the sun and relieved by sleeping/resting and intake of analgesics (paracetamol/aspirin) with occasional body itch and sweaty palms. There was no history of cough, drooling of fluid from nose, mouth or night sweats or loss of weight. There was no history of yellowness of eyes, vomiting and swelling of the limbs. No history of trauma, polydipsia, polyphagia and polyuria or whitish lesion (uraemic frost) or excoriation/cicatrization marks on the body. There was occasional intake of cassava. He was not a food choosing person but ate anything that was served. Not a strict vegetarian, not an alcoholic or smoker. No history of incontinence of urine or faeces, and phobia in him.

He was of Igbo extraction and was married to one wife with three children between them. Both parents and siblings were alive. No such complaint in his family tree. His stock in trade was business.

Examination in him revealed a middle -age man, anxious

looking, not pale, afebrile, anicteric, no lymphadenopathy, no pigmentation or whitish lesion (uraemic frost) on the body, no excoriation, no finger clubbing, no oedema but movement of legs at rest which stopped on moving about. The restless legs could not be stopped voluntarily. Fine and deep touch were preserved. Position and vibration sense were also preserved. The chest was clear and no murmur was heard. There was no gibbus on palpation of vertebral column and no musculoskeletal system abnormality. Oriented in time, place and person. Insight, judgement, memory (long and short term), attention, arithmetic, abstract thinking were all normal.

RBS = 10 mmol/l, BP = 120/80 mmHg, PR = 84 beats/min. PCV = 42%. FBC microscopy and the differential was normal, RVD - negative, S/E/U/CR normal, LFT was normal, Cxray (PA) - was normal. X ray of lumbosacral (AP and lateral) was normal.

A tentative diagnosis of Ekboms syndrome was made in the patient to exclude a background anxiety disorder in him. Patient was placed on Amitryptiline of 75mg nocte for a duration of one year. I followed him up initially at an interval of two weeks' follow up visit with mild improvement, was latter placed on two (2) months' follow up visit and subsequently placed on four (4) months' follow up visit. At the last visit a year after presentation; the patient had marked improvement with mild movement of the legs at rest and occasional paraesthesia.

DISCUSSION

The reported cases of Ekboms syndrome was made promptly because both history and examination revealed nothing that could point to a systemic ailment such as Tuberculosis since chest and vertebral column X rays were normal. ESR and FBC were also normal. There was no history of cough, smoking and the chest findings were normal; ruling out Tuberculosis or Chronic Obstructive Pulmonary Disorder (COPD). The absence of history of jaundice at any time, swelling of the body (legs, abdomen or face), generalized itching, normal liver size and a normal LFT ruled out any form of liver disease. The absence of pain, tenderness, swelling of joints, nodules, Heberdens or Bouchards nodes, deformity of interphalangeal joints of the fingers/toes-Swan-neck, Boutonniere, Square hands, Ulnar/Radial deviations ruled out any suspicion of Rheumatoid arthritis. The FBC microscopy were both normal and so ruled out any form of Vit. B₁₂, Iron deficiency and folate deficiency as one would have expected to see: microcytes, megaloblasts, macrocytes, anisococytes or target cells nevertheless patients were not pale and no koilonychia in them. There was no history of exuding fluid/catarrh from the nose, headache or facial tenderness which ruled out sinusitis or dysphagia in them ruling out any form of Patterson-Kelly

Syndrome. Following a normal S/E/U/Cr in both patients and no swelling of legs, abdomen or face, whitish lesion (uraemic frost) on the body, no change in urinary output ruling out any form of renal failure in them. There was no polydipsia, polyphagia, polyuria, nocturia, postural dizziness, vomiting, gustatory sweating or allodynia and a normal RBS in both ruling out diabetes mellitus or its complication. Since they were both non-alcoholics, that nullified the issue of alcoholic neuropathy. Varicose veins was ruled out in the absence of distended leg veins and pedal oedema. No history of palpitation, sweaty palms, diarrhoea/constipation, tremors, weight loss/gain, change in appetite, increased sensitivity to cold and a normal hair texture, reflexes and pulse rates in both ruling out any form of thyroid disease. No area of chronic muscular/joint pain and point tenderness or underlining psychological disturbance ruling out fibromyalgia. No tremors, dyskinesia, slurred speech, change in tonicidity across joints(cog-wheel, lead pipe)ruling out parkinsonism. No history of gasping for breath at the onset/during sleep ruling out sleep apnoea syndrome. No history of dryness of the mouth, non-tearing of the eyes, systemic abnormality-gastrointestinal, urogenital, respiratory system ruling out any form of autoimmune disease(ceeliac disease/sjogrens syndrome). Akathisia was borne in mind but ruled out since no history of psychosis, was never on any antipsychotic drug and an attempt to stop the restless legs voluntarily was abortive and they rarely experienced the restless legs during sleep nor worsened during stress. Both patients take cassava but because of lack of instrument to actually monitor the level of cyanide in the blood it was not done. Also the amount of magnesium ion in the serum could not be done because of lack of instrument.

CONCLUSION

(A) Ekboms Syndrome is made following - (1) compulsive irresistible movement of the legs at rest (2) stoppage of the movement of the legs on walking (3) loss of tingling sensation (paraesthesia) on walking (4) tingling sensation in the limbs prior to restless legs at rest. (B) Ekboms Syndrome is present in our environment and does not require much investigative tools to make the diagnosis but a high index of clinical suspicion is mostly required. (D) That Amitriptylline could be used as in these index cases if other drugs like L-dopa, Benzodiazepines, bromocriptine, pergolide or opiates(codeine, propoxyphene or oxycodine) are inaccessible or unaffordable by the patient. (C) The much improvement noticed in the symptoms of the second patient-headache, chest pain and the restless legs with probable background anxiety disorder with the use of Amitriptylline could imply that restless legs could be a feature of Chronic anxiety disorder (neurosis) since it is used as an anxiolytic.

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