Early Physiotherapy Intervention of a Patient with Acute Intermittent Porphyria-A Single Case Study

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Abstract

Background: The Porphyrias are metabolic disorders, each resulting from the deficiency of a specific enzyme in the heme biosynthetic pathway. Acute intermittent porphyria (AIP) is an autosomal dominant inborn error characterized by decreased activity of porphobilinogen (PBG) deaminase leading to increased levels of haem precursors, namely amino levulinic acid (ALA) and PBG. The major manifestations of theacute hepatic porphyrias are neurologic- neuropathic abdominal pain, peripheral motor neuropathy, and mental disturbanceswith attacks often precipitated by steroid, hormones, certain drugs, and nutrition influences.

Purpose: Intensive care measures and a multidisciplinary therapeutic approach are essentialbut thereis less documented evidence are available.If this aspect is incorporated early in the rehabilitation programme, then the functional recovery in such will be better.so the aim of the study is to find how Early Physiotherapy intervention using Comprehensive need based Approach to improve the functional status of porphyria patient.

Key Points of Case: A 17 years old male patient wasdiagnosed with acute intermittent Porphyria. Having the history of abdominal pain since last 3 months with family history of AIP which was diagnosed very late. A detailed Physical and Neurological examination of this patient was taken. Along with medical treatment, early physiotherapy intervention using comprehensive need base approach was being given to the patient. The Barthel Index was taken to assess the functional status of the patient and Berg Balance Scale was taken to assess balance on admission and subsequent follow-ups. Post-intervention follow-ups were taken at 6 weeks and 12 weeks period.

Conclusion: A clinically significant improvement was found on Barthel Index and Berg Balance Scale. Early Physiotherapy intervention using a Comprehensive need based Approach is highly recommended to improve the functional status of a patient with porphyria

Keywords: *Acute intermittent porphyria, Barthel Index, Berge Balance Scale, Early physiotherapy intervention.*

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Introduction

The porphyria is arare inherited group of metabolicdisorders, occurs due to the deficiency of a specific enzyme in the heme biosynthetic pathway. These enzyme deficiencies are inherited as autosomal dominant or recessive traits.[1]

Its prevalence is 2-3 cases per 100000 persons per year. Though it is not much reported in India, but the Prevalence rate is high inthe communities of western Rajasthan like Kumar and Maheshwari.2, [3]

There are mainly two types of porphyria hepaticor *erythropoietic* classified on the basis of primary site of overproduction and accumulation of the porphyrin precursoror porphyrin, in the heme biosynthesis and may have overlapping features also. [1]

Acute intermittent porphyria is the second most common form of porphyria characterized by a deficiency of the porphobilinogen deaminaseenzymewith autosomaldominant traits.[4, 5]

The main Manifestations of AIP are neurovisceral symptoms such as pain in abdomen (85-95%) which is severe and poorly localized, vomiting (50%), constipation (50%), peripheral neuropathy (42-68%), seizures (10-16%), delirium, coma and depression. Autonomic disturbances may manifest as urinary retention, paralytic ileus, restlessness, tremor, excessive sweating, tachycardia, and labile.Complications like bradycardia and sudden death have also been reported.[6, 7]

An attack of acute intermittent porphyria may be precipitated by one of the "four Ms": medication, menstruation, malnutrition, mala-Dies.Some drugs are absolutely contraindicated during acute porphyria so patients are recommended to wear an 'alert bracelet' or other identification at all times.So, in case of emergencies the identification of patient becomes easy for health care professionals.[8]

Diagnosis of AIP is confirmed by detection of porphyrin or porphyrin precursors in freshly voided urine.Classic burgundy red discoloration of long stored urine is also a clue.Quantitative measurements of PBG and ALA in urine or erythrocyte HMBS enzyme test are more reliable confirmatory tests. [9]

Haemin therapy is the only licensed therapy for symptomatic patients with acute porphyria. Removal of precipitating factors, treatment of underlying infection, a carbohydrate diet. I.V. dextrose in high doses (300-500 g/day) blocks induction of the enzyme and prevents accumulation of precursors. Heme acts by depressing the ALA synthetase enzyme. The symptoms improve readily on heme therapy generally within 24 hours. [10]

Various Physiotherapy approaches to rehabilitation have been developed to enhance motor recovery and to achieve functional independence in patients. The theoretical background and treatment strategies of the various approaches are very different. Thus, need based comprehensive approaches could be used to enhance the functional independence in the patient.

To facilitate early and better prognosis of AIP; medical management should be continued along with early interventions using need-based approaches of physiotherapy.

Case description:

History and examinations:A 17-year-old male patient with the family history of AIP complaints of abdominal pain since last 3 months for which he was hospitalized for 4-5 days. The patient experienced 1-2 episodes of the same every month. So, he was again hospitalized where all investigations were done. USG, Endoscopy, Blood and urine tests were done. On the basis of investigations, he was diagnosed with AIP, and was treated symptomatically. Later patient experienced abdominal pain and had an episode of convulsion for which he was taken to Emergency department and treated.

Investigatory findings:

Positive: PBG (Porphobilinogen)&- 5-ALA (Amino levulinic Acid)

MRI: Focal Bilateral symmetrical non-enhancing abnormal signal intensity lesions involving Bilateralcapsulo-ganglionic regions , Bilateral temporal & frontal lobes with leuko-malacic cystic change involving external capsule, internal capsule & sub-cortical white matter of temporal lobes on both sides.

On examination: Respiratory Rate was 20/min and regular, Heart Rate was 100/min and Blood Pressor-100/70 mmHg, Cold hands & feet with frequent episodes of perspiration.

- Level of consciousness: vegetative state.
- Pathological reflex: Tonic Labyrinthine Reflex

- Supine dominant.

· Postural deviation: Right scoliosis at thoraco-lumbar region.

· Generalized wasting of whole body.

· Tone in the muscles appeared fluctuating with deep tendon reflexes brisk and Babinski's sign - positive.

· Ankle Clonus present and Choreo–athetoid type of involuntary movement was present.

· Muscle twitching present at multiple muscle groups of body.

Description of outcomes: The Barthel Index was taken to assess the functional status of the patient. The Barthal Index hasan excellent reliability, validity, and sensitivity.[11]and Berg Balance Scale was taken to assess balance. It has high (r=0.95) intrarater and inter-rater reliability.[12, 13]on admission and subsequent follow-ups. Post-intervention follow-ups were taken at 6 weeks and 12 weeks period.

Description of Intervention:

Physiotherapy interventionwas given to this patient by using differentcomprehensive need based approaches–

Ø Conductive education approach- Family & parental education &counseling regarding relapes and medication.

Ø Neuro developmental therapy–Proper positioning -To prevent abnormal posture, abnormal reflex patterns, contractures & deformity, bedsores and promote development of correct joint alignment, synergy & comfort.

Ø For prevention of secondary complications like Respiratory complications – Patient is given postural drainage followed by suctioning and good Breathing exercises,for Deep vain thrombosis prevention-passive movements of lower limb. For prevention of Bedsores &other skin eruptions – advice for maintaining good hygiene and frequent change of positioning and use of water bed.for prevention of Contractures & deformities –full range passive movements and use Night splint was adviced.

Ø Rood's Approach-Improve arousal through sensory stimulation.Multisensory stimulation was used through auditory,olfactory , gustatory ,visual , tactile , kinesthetic & vestibular stimulations.

Ø Early transition to sitting postures was given as soon as patient was medically stable.

Ø Motor Relearning programme of car and shepherd- Patient is given Task – oriented activities to achieve functional activities

Ø Dietary advices given infirm of high carbohydrate.

Ø Advices on discharge –To do exercises regularly in correct manner, Proper positioning, eye care, oral care , catheter care ,trecheostomy care and maintain proper hygiene.

Table -1 Summary of values on BI AND BBS

	Barthal Index	Bearg Balance Scale
ON ADMISSION	0	0
AT 6 WEEK	70	35
AT 12 WEEK	100	56

Discussion

Primary aim of the study was to see how Early Physiotherapy intervention using Comprehensive needbased Approacheshelp in improving functional status of porphyria patient. Adetailed Physical and Neurological examination of a 17 years old male was taken The Barthel Index and BBSwas taken on

admission and subsequent follow-ups. Post-intervention follow-ups were taken at 6 weeks and 12 weeks period. A clinically significant improvement was found on Barthel Index and BBS

The possible recovery might be because of hemin therapy because Albumin-bound heme rapidly enters hepatocytes, increasing amount of functional cytochromes also increase amount of functional tryptophan pyrrolase and to replete the heme pool that regulates activity of ALA Synthesis. [14]Another study provides strong evidence that prophylactic heme therapy, through a multi-disciplinary approach, decreases the incidence of acute attacks, decreases health care costs and leads to better patient satisfaction and quality of life.[15]

Different therapeutic approaches have been used to enhance the functional recovery of patients

1) Rood's Approach-Improvement in arousal level could be due to the use of Rood's approach.

Based on available literature, Facilitation or inhibition of proprioceptors, exteroceptors, and vestibular stimulation excited the cortical level and give motor recovery. Autonomic nervous system stimulation, another component of Rood's approach can also stimulate the motor activity of vital organs as well as the skeletal muscles.[16]

2) Bobath Approach- Postural correction in patient was might be because of using Bobath approach in form of proper positioning during sitting, standing and lying as Intervention strategies and techniques for Bobath consist of therapeutic handling, facilitation, and activation of key points of control. Which was taken care during every exercise. Therapeutic handling is used in order to influence the quality of the patients' movements and incorporates both facilitation and inhibition.[17,18]

3) Motor relearning Approach -The motor relearning approach promotes the regaining of normal motor Skills through task-oriented practice with appropriate feedback and the active participation of the patients.This therapy increases the functional use of the neurologically weaker extremity through massed practice of functional activity with task performance.[19-21]

4) Conductive education is built on the assumption that the damage to the central nervous system which causes motor dysfunction can be overcome by using specialized learning strategies and that the nervous system can generate new neural connections. Education is designed to teach individuals how to complete daily tasks such as reading, eating or speaking in practical situations. The situations, be it at home in an educational setting, present opportunities for a patient to learn in real-world environments.[22]

Conclusion

A clinically significant improvement was found on Barthel Index and Berg Balance Scale. Early Physiotherapy intervention using a Comprehensive need based Approach is highly recommended to improve the functional status of a patient with porphyria

Ethical Clearance –Taken from institutional advisory board.

Consent: Written informed consent was taken from the patient.

Conflict of Interest: Nil

Source of Funding: Self

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