Psychiatric symptoms as manifestation of homocystinuria: case report

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Objective

Our main objective is to re-enforce the importance of excluding medical diagnosis in patients with psychiatric syndromes

Materials and methods

A non systematic literature search was performed in *pubmed*, *medline* and *uptodate*, using the key words “homocystinuria and psychiatry” The patient clinical record was reviewed

Homocystinuria

Homocystinuria is a disorder of methionine metabolism, leading to an abnormal accumulation of homocysteine and its metabolites (homocystine, homocysteinecysteine complex, and others) in blood and urine.

Homocystinuria is an autosomal recessively inherited defect in one of 3 possible enzymes.

- Homocystinuria is associated with the following physical findings:
- Downward dislocation of lens (ectopia lentis)
 - Marfanoid habitus
 - Pes excavatum, pes carinatum, and genu valgum
 - Mental retardation
 - Signs and symptoms of strokes in any vascular distribution: hemiplegia, aphasia, ataxia, and pseudobulbar paralsy

Homocystinuria I	cystathionine Bsynthase (CBS) enzyme	pyridoxal 5phosphat
Homocystinuria ii	methylcobalamin synthesis	methylcobalamin
Homocystinuria Iii	methylene tetrahydrofolate reductase (MTHFR)	folate

Case Report

MAR 19

- Obsessional thoughts
- “need to prove that everything is fine”
- Anxiety

1 MAY 19

- Obsessional thoughts
- “Afraid of forgetting things”
- “Afraid of saying something wrong”
- Anxiety
- Trouble sleeping

13 MAY 19

- Obsessional thoughts
- afraid of harm someone”
- “afraid of losing control”,
- Compulsive behavior
- verification behavior about confirming if he did something wrong
- Trouble sleeping
- Anxiety
- Behavior alterations
- Fluctuating leveles of orientation

Acute psychiatry unit

13 MAY 19

- **Homocystein: high (821 µmol/l)**
- Haloperidol 2mg 3id
- Cyanocobalamin 0,2mg 2id ;
- Pyridoxine 200mg 2id ;
- Thiamine 100mg 2id
- Folic acid 5mg id

30 MAY 19

- some thoughts/worries about work and verification behavior about confirming if he did something wrong
- Haloperidol 1mg 3id
- Cyanocobalamin 0,2mg 2id ;
- Pyridoxine 200mg 2id ;
- Thiamine 100mg 2id
- Folic acid 5mg id

Discharged

Follow up

14 JUN 19

- Assymptomatic
- Homocysteine (362,9 µmol/l)
- Cyanocobalamin 0,2mg 2id ;
- Pyridoxine 200mg 2id ;
- Thiamine 100mg 2id
- Folic Acid 5mg id

13 NOV 19

- Assymptomatic
- Homocysteine (< 15µmol/l)
- Cyanocobalamin 1mg
- Pyridoxine 300mg 3id
- Folic Acid 5mg id

Homocystinuria and Psychiatry

- Abbott et al 1987
- Homocystinuria commonly affects the central nervous system, mental retardition, seizures and strok
- 63 persons with homocystinuria were availed
 - psychiatric disturbance
 - intelligence
 - evidence of other central nervous system problems
 - responsiveness to vitamina B6 (pyridoxine)
- 51% were clinically significant psychiatric disorders
 - 10% depression
 - 17% chronic disorders of behaviour
 - 5 % obsessive-compulsive disorder
 - 19 % personality disorders

L. Stewart et al 1999

- female
- 31 years old
- Homocystinuria (age 22)
- delirium and inappropriate and labile affect

Ryan at al 2002

- male
- 17 years old
- No previous diagnosis of homocystinuria
- auditory and visual hallucinations and marked paranoia

Rahman T et al 2014

- female
- 42 years old
- Homocystinuria (age 12)
- Capgras delusion (belefs that neighbors were spying on her)

T. Abreu et al 2016

- male
- 22 years old
- Homocystinuria (age 4)
- anxiety and behavioral changes, as difficulty in controlling impulses

Conclusions

Despite the fact that psychiatric symptoms are frequent in homocystinuria, OCD-like symptoms are not that common. We underline the importance of excluding medical history in all cases, especially when presentation is atypical or abrupt or in treatment resistant cases.