orphanaesthesia

Anaesthesia recommendations for patients suffering from

Biotinidase deficiency

Disease name: Biotinidase deficiency

ICD 10: E53.8

Synonyms: Late-Onset Biotin-aesponsive Multiple Carboxylase Deficiency, Late-Onset Multiple Carboxylase Deficiency

Disease summary: Biotinidase deficiency (BD), biotin metabolism disorder, was first described in 1982 [1]. It is inherited as an autosomal recessive trait. The incidence of BD in the world is approximately 1/60.000 newborns [1]. Clinical manifestations include neurological abnormalities (seizures, ataxia, hypotonia, developmental delay, hearing loss and vision problems like optic atrophy), dermatological abnormalities (seborrheic dermatitis, alopecia, skin rash, conjunctivitis, candidiasis, hair loss), neuromuscular abnormalities (motor limb weakness, spastic paresis, myelopathy), metabolic abnormalities (ketolactic acidosis, organic aciduria, hyperammonemia) [1-6]. Besides, respiratory problems (apnoea, dyspnoea, tachyponea, laryngeal stridor) and immune deficiency findings (prolonged or recurrent viral/fungal infections) are associated with BD [1,3,4]. Hypotonia and seizures are the most common clinical features [4,7].

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Disease summary

Treatment with 5-10 mg of oral biotin per day results rapidly in clinical and biochemical improvement. However, once vision problems, hearing loss, and developmental delay occur, these problems are usually irreversible even if the child is on biotin therapy [4]. Moreover, BD can lead to coma and death when the child if not treated [8]. In some children, especially after puberty, biotin dose is increased from 10 mg per day to 20 mg per day. A child with profound BD has less than 10% of mean normal serum biotinidase activity, whereas a child with partial BD has 10%-30% of mean normal serum biotinidase activity. BD can be identified by newborn screening. BD gene is located on 3p25.1 of chromosomal locus [4].

Age of clinic presentation varies from 1 week to 12 years or adulthood [9,10]. Two asymptomatic adult with profound BD have been reported [11]. As BD is associated with VACTERL syndrome, annular pancreas and vascular ring malformation was reported in the literature [12,13], so they are almost certainly coincidental.

Typical surgery

Auditory system measured by tympanometry, behavioural audiometry, otoacoustic emissions (OAE), and auditory brainstem responses (ABRs) is reviewed by an expert audiologist. Auditory neuropathy or dyssynchrony have been reported in BD [14]. If hearing loss exists, 6 months use of hearing aid must be fitted with auditory rehabilitation. After then, cochlear implantation surgery may be possible in patients with severe hearing loss.

Type of anaesthesia

General or regional anaesthesia may be applied. However, during general anaesthesia in patients with hypotonia, a possibility of long-lasting neuromuscular blockade (NMB) should be kept in mind [15].

Without contraindication, regional anaesthesia may be preferred in which possible.

Necessary additional diagnostic procedures (preoperative)

Basically, if an individual with BD is being treated with biotin, they should be metabolically and immunologically in normal homeostasis and react like any normal, unaffected individuals. If they are compromised by the disorder before they were diagnosed or treated, then they may have irreversible features; however, once on biotin, they too should be biochemically stable.

Neurological symptoms (seizures, hypotonia, etc.) are reviewed by paediatric neurology. Electroencephalography during sleep or under sedation is preferred in patients that have seizures over the last six months. Motor limb weakness are assessed by electromyography is recommended.

These children are tended to upper respiratory tract infection by especially viral agents due to immune deficiency. Because of this, these children who had symptoms were reviewed by paediatric consultant.

Preoperative analysing arterial blood gas is recommended for evaluating metabolic status.

Particular preparation for airway management

There are no reports.

Particular preparation for transfusion or administration of blood products

There are no reports.

Particular preparation for anticoagulation

There are no reports.

Particular precautions for positioning, transport or mobilisation

There are no reports.

Probable interaction between anaesthetic agents and patient's long term medication

Interacting between biotin and any drugs is not reported. However, the antiepileptic drugs (AEDs) and anaesthetic drugs may interact. Some AEDs may affect biotin absorption. The AEDs affect hepatic enzymes that may change metabolism of anaesthetic drugs. Therefore, the resistance to opioids and NMB agents can be occurred. In addition to the adverse effects of AEDs, such as sedation, drowsiness and somnolence should be kept in mind [16].

Anaesthesiologic procedure

It is well known that cases should be individualized. Does not necessarily deal with an individual with BD.

Premedication with metoclopramide and histamine-2 receptor antagonists or proton pump inhibitors per intravenously at 30 minutes before surgical procedure for risk of gastric aspiration is recommended. Benzodiazepines, especially midazolam, which is a short acting benzodiazepine, can be used for sedation and preventive effect of the new epileptogenic activity [17,18]. However, preoperative deep sedation should be avoided.

In these patients, there is no contraindication for general anaesthesia, but there are some special properties. The new epileptic activity, acidosis, long-lasting NMB and malignant hyperthermia could be occurred.

In minor surgical procedure and if no needed neuromuscular blockade, laryngeal mask airway (LMA) anaesthesia using low dose propofol and opioids, such as alfentanil, remifentanil and fentanyl, without NMB agents is recommended. Moreover, patient's ventilation is maintained spontaneously during the surgery [17].

In major surgical procedure, total intravenous anaesthesia (TIVA) is recommended for general anaesthesia. At induction of anaesthesia, propofol and opioids can be used. At maintenance of anaesthesia is provided propofol and remifentanil infusion. TIVA with propofol and remifentanil was preferred as a safe technique in children with hypotonia or seizures in the literature [19-21].

At induction of anaesthesia, thiopental had a property of inhibiting epileptic activity is used especially in patient with seizures. Etomidate should be avoided because of its myoclonic movements. The use of ketamine is controversial in patients with seizures [18].

With regards to the use of opioids, the high dose of opioids is not recommended in hypotonic children [17,20].

Sevoflurane, one of inhalation agent, is commonly used for induction anaesthesia, if intravenous cannulation is not provided. Sevoflurane was associated with abnormal epileptiform activity during induction of anaesthesia [22]. Because of this, it could be used minimal concentration. Additionally, sevoflurane combination with nitrous oxide is recommended to avoid its inducing epileptiform activity. By using adjunctive drugs (opioids, nitrous oxide, benzodiazepines, etc.), inhalation agents should be used below 1.5 MAC (minimum alveolar anaesthetic concentration) during maintenance of anaesthesia [18]. Nitrous oxide tends to inhibit seizure-like activity [16].

Because hypotonia is a common pathology in these children, neuromuscular block agents should be avoided, if possible. However, if needed, rocuronium is preferred initially [19]. Steroidal NMB agents, such as especially rocuronium, vecuronium and pancuronium, are recommended because these agents may be reversed by sugammadex. Because of risk of hyperkalaemia, malignant hyperthermia and epileptiform activity, succinylcholine should be avoided [23].

Regional anaesthesia is preferred by avoiding general anaesthesia and its perioperative negative effects. Additionally, regional anaesthesia has better postoperative analgesia than general anaesthesia. Central neuroaxial and peripheral nerve blocks may be applied under sedation, if possible. With regards to these techniques' difficulties, skin lesions may be existed on applying zone and appropriate positioning of patient may be difficult owing to neuromuscular abnormalities.

Particular or additional monitoring

The minor or short surgeries are not needed additional monitoring if the child is asymptomatic and has normal routine tests. However, in major surgeries, arterial cannulation for observing invasive blood pressure and analysing arterial blood gas should be provided. Central vena cannulation for fluid replacement and blood or blood product transfusion if needed should be provided. Body temperature and neuromuscular blockade monitoring is recommended [15].

Possible complications

New epileptic activity, acidosis, respiratory problems associated with/without long-lasting NMB, malignant hyperthermia may be occurred in patients with the presence of hypotonia or seizures [15,16,23].

Postoperative care

Postoperative care is depended on the preoperative condition of patients and surgical procedure. Postoperative care unit is not mandatory for minor or short surgeries and cases which applied regional anaesthesia. However, close observation and supporting mechanical ventilation in postoperative care unit is needed in case of postoperative respiratory problems due to existing hypotonia.

Information about emergency-like situations / Differential diagnostics

Caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the diseases, e.g.:

Respiratory problems, hyperthermia and skin rash may be caused by illness or anaesthetic procedure.

Ambulatory anaesthesia

There are no reports.

Obstetrical anaesthesia

There are no reports.

Literature and internet links

1. Wolf B, Grier RE, Secor McVoy JR, Heard GS. Biotinidase deficiency: a novel vitamin recycling defect. J Inherit Metab Dis 1985;8:53-8.
2. Komur M, Okuyaz C, Ezgu F, Atici A. A girl with spastic tetraparesis associated with biotinidase deficiency. Eur J Paediatr Neurol 2011;15:551-3
3. Kalkanoglu HS, Dursun A, Tokatli A, Coskun T, Karasimav D, Topaloglu H. A boy with spastic paraparesis and dyspnea. J Child Neurol 2004;19:397-8
4. Wolf B. Biotinidase Deficiency. In : GeneReviews(Internet). Eds:Pagon RA, Adam MP, Ardinger HH, et al. Seattle (WA): University of Washington, Seattle 2013
5. Wiznitzer M, Bangert BA. Biotinidase deficiency: clinical and MRI findings consistent with myelopathy. Pediatr Neurol 2003;29:56-8
6. Chedrawi AK, Ali A, Al Hassnan ZN, Faiyaz-Ul-Haque M, Wolf B. Profound biotinidase deficiency in a child with predominantly spinal cord disease. J Child Neurol. 2008;23:1043-8
7. Wolf B. The neurology of biotinidase deficiency. Mol Genet Metab 2011;104:27-34
8. Baykal T, Hüner G, Sarbat G, Demirkol M. Incidence of biotinidase deficiency in Turkish newborns. Acta Paediatr 1998;87:1102-3
9. Venkataraman V, Balaji P, Panigrahi D, Jamal R. Biotinidase deficiency in childhood. Neurol India 2013; 61:411-3
10. Casado de Frias E, Campos-Castello J, Careaga Maldonado J, Perez Cerda C. Biotinidase deficiency: Result of treatment with biotin from age 12 years. Eur J Paediatr Neurol 1997; 1:173-6
11. Wolf B, Norrgard K, Pomponio RJ, Mock DM, McVoy JR, Fleischhauer K, Shapiro S, Blitzer MG, Hymes J. Profound biotinidase deficiency in two asymptomatic adults. Am J Med Genet 1997; 73:5-9
12. Sezer R, Aydemir G, Bozaykut A, Paketci C, Aydınoz S. VACTERL association: a new case with biotinidase deficiency and annular pancreas. Ren Fail 2012;34:123-5
13. Gonzalez-Salazar F, Gerardo-Aviles GJ, Jacobo RS, Vargas-Camacho G. [Biotinidase deficiency and vascular ring malformation: case report]. Arch Argent Pediatr 2014;112:217-21
14. Talebi H, Yaghini O. Auditory Neuropathy/Dyssynchrony in Biotinidase Deficiency. J Audiol Otol 2016;20:53-4
15. Rakow H. Anesthesia recommendations for patients suffering from Prader-Willi syndrome. Orphan Anesthesia 2012;1-8. http://www.orphananesthesia.eu/en/rare-diseases/published-guidelines/doc_download/96-prader-willi-syndrome.html
16. Kofke WA. Anesthetic management of the patient with epilepsy or prior seizures. Curr Opin Anaesthesiol 2010; 23:391-9
17. Goktas U, Cegin MB, Kati I, Palabiyik O. Management of anesthesia in biotinidase deficiency. J Anaesthesiol Clin Pharmacol 2014;30:126
18. Voss LJ, Sleigh JW, Barnard JP, Kirsch HE. The howling cortex: seizures and general anesthetic drugs. Anesth Analg. 2008;107:1689-703
19. Tan SH, Ng VH. Anaesthesia for a child with adrenoleukodystrophy: A case report and review of the literature. Indian J Anaesth 2014;58:63-5
20. Buntenbroich S, Dullenkopf A. Total intravenous anesthesia in a patient with Joubert-Boltshauser syndrome. Paediatr Anaesth 2013;23:204-5
21. Kocamanoglu IS, Sarihasan E. Anesthetic management of a pediatric patient with Leigh Syndrome. Rev Bras Anestesiol 2013;63:220-2
22. Constant I, Seeman R, Murat I. Sevoflurane and epileptiform EEG changes. Pediatr Anesth 2005;15:266-74
23. Saettele AK, Sharma A, Murray DJ. Case scenario: Hypotonia in infancy: anesthetic dilemma. Anesthesiology 2013;119:443-6.

Last date of modification: August 2016

This guideline has been prepared by:

Author
Onur Palabiyik, Anaesthesiologist, Sakarya University Training and Research Hospital Clinic of Anaesthesiology, Sakarya, Turkey
mdpalabiyikonur@yahoo.com

Peer revision 1
Berry Wolf, Department of Medical Genetics, Henry Ford Hospital, Detroit, Michigan, USA
bwolf1@hfhs.org

Peer revision 2
Hossein Talebi, Department of Audiology, Faculty of Rehabilitation, Communication Disorders Research Center, Isfahan University of Medical Sciences, Hazarjarib Avenue, Isfahan, Iran
ht6023@gmail.com

Please note that this guideline has not been reviewed by two anaesthesiologists, but by two disease experts instead.