orphanaesthesia

Anaesthesia recommendations for

Biotinidase deficiency

Disease name: Biotinidase deficiency

ICD 10: E53.8

Synonyms: Late-onset biotin-responsive 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 new-borns [1]. Clinical manifestations include neurological abnormalities (seizures, ataxia, hypotonia, developmental delay, hearing loss and vision problems like optic atrophy), dermatological abnormalities (seborrhoeic dermatitis, alopecia, skin rash, conjunctivitis, candidiasis, hair loss), neuromuscular abnormalities (motor limb weakness, spastic paresis, myelopathy), metabolic abnormalities (ketolactic acidosis, organic aciduria, hyperammonaemia) [1-6]. Besides, respiratory problems (apnoea, dyspnoea, tachypnoea, 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].

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 if the child is not treated [8]. In some children, especially after puberty, the biotin dose is increased from 10mg per day to 20mg 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 new-born screening. The BD gene is located on chromosome 3, gene locus 3p25.1 [4].

The age of clinic presentation varies from 1 week to 12 years or adulthood [9,10]. Two asymptomatic adults 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.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

Typical surgery

Examination of the auditory system by applying tympanometry, behavioural audiometry, otoacoustic emissions (OAE), and auditory brainstem responses (ABRs); Cochlear implantation in some cases.

Type of anaesthesia

General or regional anaesthesia may be possible.

In case of general anaesthesia, both TIVA and the use of volatile anaesthetics have been reported. Some authors report of long-lasting neuromuscular blockade (NMB) due to existing muscular hypotonia [15].

Without contraindication, regional anaesthesia may be preferred if possible and tolerated by the patient.

Necessary additional pre-operative testing (beside standard care)

Basically, if individuals with BD are treated with biotin, they should be metabolically and immunologically in normal homoeostasis and react like any 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.

Review of neurological symptoms (seizures, hypotonia, etc.) is recommended if clinically relevant.

These children are susceptible to upper respiratory tract infections especially by viral agents due to immune deficiency. In case of clinical symptoms, children should be evaluated by a paediatrician.

In case of clinical symptoms or history of metabolic disturbances, pre-operative arterial blood gas analysis is recommended to evaluate the patient's 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, transportation and mobilisation

There are no reports.

Interactions of chronic disease and anaesthesia medications

Interactions between biotin and drugs have not been reported. However, antiepileptic drugs (AEDs) and anaesthetic drugs might interact. Some AEDs might affect biotin absorption. The AEDs affect hepatic enzymes that are likely to change the metabolism of anaesthetic drugs. Therefore, resistance to opioids and NMB agents can occur. In addition to the adverse effects of AEDs, others such as sedation, drowsiness and somnolence should be kept in mind [16].

Anaesthetic procedure

In these patients, there is no contraindication for general anaesthesia. In symptomatic cases (before biotin therapy ore late biotin therapy) epilepsy, elevated risk of gastric aspiration, muscular hypotonia or metabolic disturbances (acidosis) must be acknowledged.

Intravenous premedication with metoclopramide and histamine-2 receptor antagonists or proton pump inhibitors 30 minutes before surgical procedure may be recommended in some patients because of the risk of gastric aspiration. Benzodiazepines, especially midazolam, which is a short-acting benzodiazepine, can be used for sedation [17,18]. However, pre-operative deep sedation should be avoided.

In minor surgery, the airway can be secured by a laryngeal mask using propofol and opioids, such as alfentanil, remifentanil and fentanyl, without NMB agents. Moreover, the patient's ventilation is maintained spontaneously during surgery [17].

In major surgical procedure, tracheal intubation may be preferred, Total intravenous anaesthesia (TIVA) is recommended for general anaesthesia. TIVA with propofol and remifentanil was a preferred safe technique in children with hypotonia or seizures as described in the literature [19-21].

At induction of anaesthesia, thiopental had a property of inhibiting epileptic activity and 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].

Opioids can be used safely if the dose is adapted to muscular hypotonia.

Sevoflurane, an inhalation agent, can be used for the induction of anaesthesia if intravenous cannulation cannot be established. Sevoflurane was associated with abnormal epileptiform activity during induction of anaesthesia [22]. Because of this, it should be used in minimal concentrations. Additionally, sevoflurane combined with nitrous oxide is recommended to prevent its epileptiform activity [16].

Because hypotonia is a common pathology in these children, neuromuscular blocking agents should be avoided, if possible. However, if needed, rocuronium is recommended [19]. If necessary, rocuronium can be reversed by sugammadex. Because of the risk of hyperkalaemia, succinylcholine should be avoided [23].

Regional anaesthesia is preferred when general anaesthesia shall be avoided. In addition, regional anaesthesia induces postoperative analgesia. Central neuroaxial and peripheral nerve blocks may be applied under sedation, if possible. Considering the difficulties of these techniques, skin lesions may prevail in the application area and the appropriate positioning of the patient may also be difficult owing to neuromuscular abnormalities.

Particular or additional monitoring

Minor or short surgeries will not need additional monitoring if the child is asymptomatic and is undergoing normal routine tests. However, in major surgeries, arterial cannulation for observing invasive blood pressure and analysing arterial blood gas should be provided. Central venous cannulation for fluid replacement or a transfusion of blood or blood products, 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 occur in patients with hypotonia or seizures [15,16,23].

Post-operative care

Postoperative care is dependent on the pre-operative condition of patients and the surgical procedure. Post-operative care is not mandatory for minor or short surgeries and cases in which regional anaesthesia has been applied. However, close observation and supporting mechanical ventilation in the post-operative care unit is needed in case of post-operative respiratory problems arising due to existing hypotonia.

Disease-related acute problems and effect on anaesthesia and recovery

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.

References

1. Wolf B, Grier RE, Secor McVoy JR, Heard GS. Biotinidase deficiency: a novel vitamin recycling defect. J Inherit Metab Dis 1985;8:53–58
2. Komur M, Okuyaz C, Ezgu F, Atici A. A girl with spastic tetraparesis associated with biotinidase deficiency. Eur J Paediatr Neurol 2011;15:551–553
3. Kalkanoglu HS, Dursun A, Tokatli A, Coskun T, Karasimav D, Topaloglu H. A boy with spastic paraparesis and dyspnea. J Child Neurol 2004;19:397–398
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;29:56–58
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;23:1043–1048
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;87:1102–1103
9. Venkataraman V, Balaji P, Panigrahi D, Jamal R. Biotinidase deficiency in childhood. Neurol India 2013; 61:411–413
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;1:173–176
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;34:123–125
13. Gonzalez-Salazar F, Gerardo-Aviles GJ, Jacobo RS, Vargas-Camacho G. [Biotinidase deficiency and vascular ring malformation: case report]. Arch Argent Pediatr 2014;112:217–221
14. Talebi H, Yaghini O. Auditory Neuropathy/Dyssynchrony in Biotinidase Deficiency. J Audiol Otol 2016;20:53–54
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; 23:391–399
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;107:1689–1703
19. Tan SH, Ng VH. Anaesthesia for a child with adrenoleukodystrophy: A case report and review of the literature. Indian J Anaesth 2014;58:63–65
20. Buntenbroich S, Dullenkopf A. Total intravenous anesthesia in a patient with Joubert-Boltshauser syndrome. Paediatr Anaesth 2013;23:204–205
21. Kocamanoglu IS, Sarihasan E. Anesthetic management of a pediatric patient with Leigh Syndrome. Rev Bras Anestesiol 2013;63:220–222
22. Constant I, Seeman R, Murat I. Sevoflurane and epileptiform EEG changes. Pediatr Anesth 2005;15:266–274
23. Saettele AK, Sharma A, Murray DJ. Case scenario: Hypotonia in infancy: anesthetic dilemma. Anesthesiology 2013;119:443–446.

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This recommendation was prepared by:

Author

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

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This recommendation was reviewed by:

Reviewer 1

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

Reviewer 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.

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