PHO Benguet Conducts Common Metabolic Disorders Forum

The Provincial Health Office (PHO) of Benguet is partnering with Regional Genetic Hospital and Medical Center (RHMC), and the Integrated Provincial Health Officer’s Office (IPHO) of Benguet in the implementation of the Common Metabolic Disorders (CMDs) Forum. The event was attended by health workers and other stakeholders who underscored the need for timely diagnosis and management of disorders such as Tay-Sachs disease, Hurler’s Syndrome, Pompe disease, etc.

Mindanao CHDs, NSC-M Hold NBS Trainings

The National Screening Center (NSC) is organizing trainings on newborn screening (NBS) for the provinces and cities of Mindanao. The trainings aim to ensure that all facility coordinators are equipped with the proper knowledge and skills in implementing the program. The trainings were facilitated by Julie An, a professional in the field of newborn screening.

NSC–Northern Luzon (NSC-NL) underwent and passed a 2019 Semi-Annual PIR

The NSC-NL started offering NBS services to Ilocos Province in 2016 and has been management, while Personal Parents' Panel, including families with affected babies, is also active in this process.

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Homocystinuria (Hcy) is caused by cystathionine β-synthase (CBS) deficiency, a disorder affecting a conserved enzyme responsible for the remethylation of homocysteine (Hcy) to methionine (Met) in the transsulfuration pathway of sulfur amino acid metabolism.

Clinical Manifestation

Children with homocystinuria may present with cognitive delay, behavioral disturbances, headaches, and retinal abnormalities. They may also exhibit skeletal abnormalities such as lens dislocation, skeletal overgrowth, and increased fracture risk.

Pathophysiology

CBS deficiency leads to increased plasma levels of Hcy. Hcy is a powerful prooxidant that can cause oxidative stress and damage to DNA, proteins, and lipids. Increased levels of Hcy can also lead to increased production of homocysteine, which can contribute to increased thrombosis and atherogenesis.

Metabolic Crises

Metabolic crises in patients with CBS deficiency can be triggered by illness, infection, surgery, and stress. These crises can cause acidosis, azotemia, and thromboembolism.

Diagnostic Tests

The diagnosis of homocystinuria is confirmed by elevated plasma total homocysteine levels. DNA analysis of CBS gene mutations can also be performed.

Management

The aim of treatment is to reduce the plasma total homocysteine levels by lowering dietary intake of methionine and cysteine, and providing vitamin supplementation. Additional treatment may include vitamin C (100 mg/day) and vitamin E (400 IU/day) if there is evidence of oxidative stress.

What to Do

- Monitor input and output strictly (q6 hours).
- Start betaine, folic acid, and vitamin B6.
- Insert IV access. Collect samples for plasma amino acids.
- Ensure patient’s airway is secure.
- If unwell and can tolerate oral intake:
  - Start D12.5% 0.3NaCl at 5-10 cc/hr.
- If unwell and cannot tolerate oral intake:
  - Start D12.5% 0.3NaCl at full maintenance. Assess patient clinically; if there is need to increase fluid, may do so up to 1.2 times maintenance.
- Monitor EKG, BP, and heart rate.
- Take care of essential metabolic needs (acidosis, azotemia).
- Inform metabolic doctor on call for further guidance regarding ongoing management.
- Monitor CBC, blood urea nitrogen, creatinine, and electrolytes.
- Monitor blood pressure and heart rate regularly.

In conclusion, early diagnosis and treatment can prevent thromboembolic events and reduce the complications brought about by increased homocysteine levels. It is crucial to have a comprehensive newborn health program for the benefit of the newborns and their families.

October 1-7, 2019. Work with us in saving Filipino newborns from mental retardation and death!}

What’s inside

1. NSC-NL Passes Reaccreditation
2. PHD Benguit Conducts Common Metabolic Disorders (IMHO-Regional) Field Test 2015 Semi-Annualinar Program
3. Malabon City, NSC-NL Hold NSN Trainings
4. NSC-SL Organizes 2nd Thalassaemia Forum
5. 3rd Annual Congress
6. In The Know Homocystinuria