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REVIEWER'S REPORT

Manuscript No.: IJAR -59148

Title: ***A CASE REPORT ON PHENYLKETONURIA:*** **EARLY INTERVENTION AND TREATMENT.**

Recommendation:
Accept as it is .....
Accept after minor revision.....
Accept after major revision .....
Do not accept (*Reasons below*) .....

Rating	Excel.	Good	Fair	Poor
Originality		Good		
Techn. Quality	Excellent			
Clarity	Excellent			
Significance	excellent			

Reviewer's ID: Dr. Sumathi

Detailed Reviewer's Report

1. **Phenylketonuria (PKU) is a rare, inherited metabolic disorder that causes a dangerous buildup of the amino acid phenylalanine in the body. Phenylalanine is a building block of protein found in many common foods and some artificial sweeteners.**
2. **People with PKU lack a functional enzyme called phenylalanine hydroxylase (PAH), which is responsible for converting phenylalanine into another amino acid, tyrosine. When this process fails, toxic levels of phenylalanine accumulate in the blood and brain, which can cause permanent brain damage and severe intellectual disabilities if left untreated.**
3. **Phenylalanine hydroxylase deficiency, commonly known as phenylketonuria (PKU), is a rare inherited metabolic disorder. It is caused by mutations in the PAH gene, which provides instructions for making the enzyme phenylalanine hydroxylase. Without this enzyme, the body cannot properly break down phenylalanine (Phe)—an essential amino acid found in protein-rich foods and certain artificial sweeteners like aspartame. This leads to a toxic**

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buildup of Phe in the blood and brain, alongside a deficiency of tyrosine.

4. **Hyperphenylalaninemia (HPA) is a medical condition characterized by abnormally elevated levels of phenylalanine (an essential amino acid) in the blood. It is the most common inherited defect of amino acid metabolism in humans. When we eat protein-rich foods, our body breaks them down into amino acids. Normally, an enzyme called phenylalanine hydroxylase (PAH) converts phenylalanine into another amino acid called tyrosine. In individuals with HPA, this process fails due to a total or partial deficiency of the PAH enzyme or its crucial cofactor, tetrahydrobiopterin (BH4).**
5. **A phenylalanine (Phe) restricted diet is a highly specialized medical diet used primarily to manage phenylketonuria (PKU), a rare genetic disorder. Individuals with PKU lack the enzyme needed to break down phenylalanine, an essential amino acid found in protein. Without management, toxic levels of Phe can build up in the blood and brain, causing severe neurological issues.**
6. **Attention-deficit/hyperactivity disorder (ADHD) is a biological, neurodevelopmental condition that alters how the brain regulates attention, impulse control, and activity levels. It is not a failure of willpower or discipline; rather, it is a structural and chemical difference in neural networks—primarily involving dopamine and norepinephrine—that affects executive function and self-regulation. Symptoms typically emerge in early childhood and frequently persist through adolescence into adulthood.**
7. **Newborn screening (NBS) is a crucial public health procedure performed on infants shortly after birth—typically between 24 and 48 hours of age—to detect serious, rare, and life-threatening genetic, metabolic, and developmental conditions before symptoms appear.**
8. **The PAH gene provides the essential genetic instructions for manufacturing an enzyme called phenylalanine hydroxylase. This enzyme plays a critical role in metabolic health by breaking down phenylalanine, an amino acid obtained through protein-rich dietary sources like meat, eggs, dairy, and certain artificial sweeteners.**
9. **A metabolic disorder occurs when abnormal chemical reactions in the human body disrupt the normal process of metabolism—the critical mechanism that converts macronutrients (proteins,**

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carbohydrates, and fats) into energy. When these pathways break down, the body may end up with too much or too little of essential substances required to sustain health.

10. **Key words are given very impressive.**
11. **Significant points are given very meaningful.**
12. **In result part can be given tables with graphs for values.**
13. **Summary points only can be included.**
14. **References should be with alphabetical order.**
15. **After those small changes good to publish in your journal.**