Assessment of Mothers Care toward their Children Having Phenylketonuria

Thesis

Submitted for Partial Fulfillment of the Requirements for Master Degree in **Pediatric Nursing**

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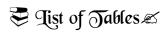


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List of Abbreviation

AA : Amino Acid

ADHD : Attention-Deficit/Hyperactivity Disorder

ASD : Autism Spectrum Disorder

BH4 : Tetrahydrobiopterin

BMI : Body Mass Index

BDI : Beck Depression Inventory Scale

DRI: Daily Requirements Intake

EEGs: Electroencephalograms

ESPKU: European Society of Phenylketonuria

and Allied Disorders

GMP: Glycomacropeptide

GTP : Guanosine Triphosphate

HPA: Hyperphenylalaninemia

Hrs: Hours

IEM: Inborn Error of Metabolism

IMDs: Inherited Metabolic Disorders

IDSA : Infectious Diseases Society of America

LNAA: Large Neutral Amino Acids

Mmol : Millimoles

MRI : Magnetic Resonance Imaging

NBS: Newborn Screening

NIH : National Institute of Health

🕏 Iist of Abbreviations 🗷

NSE: National Sleep Foundation

NPKUA: National Phenylketoneuria Association

PHE: Phenylalanine

PAH: Phenylalanine Hydroxylase

PHE : Phenylalanine Hydroxylase Enzyme

PKU: Phenylketoneuria

SD : Standard Deviation

STDs: Sexually Transmitted Disease

SPSS: Statistical Package for Social Science

S-TAI : State-Trait Anxiety Inventory

Tyr: Tyrosine

USA: United State of America

WHO: World Health Organization

Theoretical Definitions

Amino acid: Organic compounds that form the building blocks of protein. There are 20 types of amino acids (nine are "essential amino acids" which the body cannot make and must be obtained from food).

ESPKU: The European Society for Phenylketonuria and Allied Disorders is the umbrella organization of national and regional associations from about 30 countries established by parents.

Enzymes: A protein that catalyzes a biochemical reaction or change without changing its own structure or function.

Inherited metabolic diseases: A group of genetic disorders characterized by specific enzymatic defects leading to accumulation of metabolites in various tissues and organs resulting in pathologic sequels. The detection of metabolic disorder is done either by measuring the enzyme activities or detection and quantification of the abnormal metabolites by different available techniques.

Newborn screening: Is one of the first and largest population-based disease intervention programs. These programs were initially designed to detect infants with possible inborn errors of metabolism which are genetically determined and typically inherited in an autosomal recessive pattern.

Phenylalanine (**phe**): Is one of the essential amino acids. The liver uses an enzyme called phenylalanine hydroxylase (PAH) together with a cofactor (BH4) to change some of the Phe to a non-essential amino acid called tyrosine (Tyr). Tyr is not only used to build your own proteins but is also.

Phenylalanine hydroxylase: Is an enzyme the body uses to convert phenylalanine into tyrosine, which the body needs to create neurotransmitters such as epinephrine, nor epinephrine, and dopamine.

Protein: Building blocks of the body, composed of amino acids, involved in the formation of body structures and controlling the basic functions of the human body.

Genetic disease: A disease that is (partly or completely) the result of the abnormal function or expression of a gene; a disease caused by the inheritance and expression of a genetic mutation.

Hyperphenylalaninemia: presence of blood phenylalanine levels that exceed the limits of the upper reference range (2 mg/dL or 120 μmol/L) without treatment but that are below the level found in patients with phenylketonuria (PKU).

Mutation: A permanent change in the genetic material that may alter a trait or characteristic of the child, or manifest as disease, and can be transmitted to offspring.