Assessment of Mothers Care toward their Children Having Phenylketonuria

Thesis

Submitted for Partial Fulfillment of the Requirements

for Master Degree in ***Pediatric Nursing***

By

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Amira Khalil Ibrahim

List of Contents

List of Tables

List of Figures

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

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.