

NEURODEGENERATIVE
DISEASES PRESENTING IN INFANCY
AND CHILDHOOD PERIOD

ESSAY

SUBMITTED FOR PARTIAL FULLFILMENT
OF MASTER DEGREE IN PEDIATRICS

BY

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بِسْمِ اللَّهِ الرَّحْمَنِ الرَّحِيمِ



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الْأَبْدَانِ لِلَّهِ طَهَّرَ الْقُلُوبَ

To my mother

Who Self-sacrifices
for me and my brothers

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=====

"First and foremost , thanks are to GOD the beneficent and merciful " .

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7

Errata

Page	Line	Error	Correct
3	20	and	end
5	9,11	symptomes	symptoms
6	5	muscloskletal	musculoskeletal
6	18	deficalties	difficulties
6	19	predominent	predominant
7	3	atroply	atrophy
7	4	destruction myelin	destruction of myelin
10	5	definit	definite
10	11	Exagerated	Exaggerated
11	11	paraplagia	paraplegia
13	16	hepatis	hepatitis
16	20	accampnied	accompanied
17	4	interrmettent	intermittent
17	14	manefist	manifest
17	16	priviously	previously
18	1	parants	parents
19	21	promenent	prominent
20	19	frequently	frequently
20	21	disterbance	disturbance
20	22	evedent	evident
22	3	exedent	exident
23	8	faiiliure	failure
25	5,10	discrbed	described
25	11	constructures	contractures
25	19	thickning	thickening

Page	Line	Error	Correct
26	16	vascuolar	vascular
30	4,13	headiche	headache
30	14	conciuousness	consciousness
33	8	magority	majority
35	18	ultrastractural	ultrastructural
41	17	lumber	lumbar
43	14	forth	fourth
46	16	manifisted	manifested
47	12	cellulra	cellular
48	2	ultrastractural	ultrastructure
50	5	particutarly	particularly
51	4	lumber	lumbar
52	22	lumber	lumbar
53	18	rarefication	rarefaction
56	4	restrected	restricted
56	12	membrain	membrane
58	17	vescera	viscera
58	24	ultrastractural	ultrastructural
119	20	encephalomyilitis	encephalomyelitis

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ABBREVIATIONS

CNS	Central nervous system
CSF	Cerebrospinal fluid
CT	Computerized tomography
DNA	Desoxyribonucleic acid
EEG	Electroencephalogram
e.r.g.	Electroretinogram
GABA	Gamma aminobuteric acid
HHE	Hemiconvulsions, hemiplegia, and epilepsy syndrome
MLD	Metachromatic leukodystrophy
NCL	Neuronal ceroid lipofuscinosis
NF	Neurofibromatosis
PDC	Pyruvate decarboxylase
TPP	Thiamine pyrophosphate
TS	Tuberous sclerosis
TTP	Thiamine triphosphate

INTRODUCTION

The neurodegenerative disorders include a number of rare but important progressive degenerative diseases of the nervous system. The gray matter of the brain is principally affected, and in the second, the leukodystrophies or disorders of white matter; demyelination is the main feature, although there is some overlapping of these categories (Brett and Lake, 1983).

Particularly in this group of children, the art of examination and interpretation based upon the full understanding of neurologic signs at different developmental ages, associated with careful history gathering, provides a data based for the determination of disease classification. Central nervous system neoplasms, infections, trauma, vascular disorders, and intoxications may produce early findings similar to those associated with primary degenerative diseases of the central nervous system structure (Allen, 1982).

Most are inherited in an autosomal recessive fashion and show involvement of different parts of the central and sometimes peripheral nervous system with a variety of progressive deficits, especially dementia, epilepsy, blindness, ataxia, and disorders of tone and reflexes (Brett a , 1983).

No effective basic treatment is get possible in these conditions which are uniformly fatal. Diagnostic accuracy is essential so that parents may be advised of the grim prognosis and also of the genetic implications with the high risk of recurrence in later pregnancies (Brett and Lake , 1983).