Oro-dental anomalies and Gingival Biopsy as a Possible Diagnostic Tool in some Autosomal Recessive Neurodegenerative disorders.

A Thesis submitted in partial fullfillement for

Master degree in

Oral Biology

By

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٢٠٠٧

بسم الله الرحمن الرحيم

"قالوا سبحانك لا علم لنا إلا ما علمتنا إنك أنت العليم الحكيم"

صدق الله العظيم

سورة البقرة الايه- ٣٢

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To my Family,

With all love and

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List of abbreviations

BAEP	Brain auditory evoked potentials
CLN	Ceroid Lipofuscinosis, Neuronal
CT	Computerized tomography
DNA	Deoxyribonucleic acid
EEG	Electroencephalography
EM	Electron microscope
EMG	Electromyography
ER	The endoplasmic reticulum
ERG	Electroretinogram
GAN	Giant axonal neuropathy

GFAP	Glial fibrillary acidic protein
GRODs	Granular osmiophilic deposits
HSS	Hallervorden-Spatz syndrome
IF	Intermediate filaments
INAD	Infantile Neuroaxonal dystrophy
INCL	Infantile Neuronal ceroid lipofuscinosis
JNCL	Juvenile Neuronal ceroid lipofuscinosis
LINCL	Late infantile Neuronal ceroid lipofuscinosis
MBP	Myelin basic protein
MLD	Metachromatic leukodystrophy
MRI	Magnetic resonance imaging
MRI	Magnetic reasonance image
NCL	Neuronal ceroid lipofuscinosis
NCV	Nerve conduction velocity
NE	Northern epilepsy
PANK٢	Pantothenate kinase ٢ enzyme
PKAN	Pantothenate kinase-associated neurodegeneration
PLP	Proteolipid protein
RER	Rough endoplasmic reticulum
RNA	Ribonucleic acid
TEM	Transmission electron microscope
VEP	Visual evoked potentials
WD	Wilson’s disese

Introduction and Review of literature

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Structure of the Gingiva

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Gingiva is the part of the oral mucosa that surrounds the crevices of the teeth; it is firmly attached to the alveolar process and the cervical parts of the teeth~~.~~ (**Melfi and Alley, ٢٠٠٠**). ~~(**Melfi and Alley ٢٠٠٠**). The two main tissue components of the~~The oral mucosa ~~are~~ consists of a surface layer of ~~a~~ stratified squamous epithelium, called the oral epithelium, and an underlying connective tissue layer, called the lamina propria~~.~~ (**Squier and Finkelstein, ٢٠٠٣**).

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~~(**Squier and Finkelstein ٢٠٠٣**).~~

~~Several layers of cells of distinct morphologies may be recognized in the~~The stratified squamous epithelium appeared to consist of several layers including~~. These layers are~~ stratum germinativum (~~or~~ stratum basale), stratum spinosum (~~or~~ prickle cell layer), stratum granulosum (~~or~~ granular layer), and stratum corneum (~~the keratinized or~~ cornified layer)~~.~~ (**Berkovitz et al., ٢٠٠٢**).

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The ~~(**Berkovitz et al ٢٠٠٢**).~~

~~Cells~~cells of the basal layer ~~are~~ proved to be the least differentiated oral epithelial cells. They contain not only the well known organelles ~~(nuclei, mitochondria, ribosomes, endoplasmic reticulum, and Golgi complexes)~~ commonly present in the cells of other tissues but also, certain characteristic structures that identify them as epithelial cells and distinguish them from other cell types. These structures are the filamentous strands called tonofilaments and the intercellular bridges or desmosomes~~.~~

(Squier and Finkelstein, ٢٠٠٣).

~~**(Squier and Finkelstein ٢٠٠٣).**~~

The nucleus is the largest, densest, and most conspicuous organelle in the cell. ~~When stained with basic histological dyes, the chief nucleoprotein in the nucleoplasm, chromatin shows up well in groups of varying size when the cell is viewed microscopically. In an actively dividing cell, the chromatin concentrates into microscopically visible, discrete, rod-like chromosomes. The nucleus~~It stores the genetic code~~.~~ and ~~From~~ from its sequence of nucleotides in the chromatin, deoxyribonucleic acid~~,~~ (DNA) ~~or DNA~~ and ribonucleic acid ~~, or~~(-RNA) give directions for everything the cell is and will be. The fluid portion within the nucleus is the nucleoplasm which contains important proteins. The nucleus is completely surrounded by the nuclear envelope, a membrane similar to the cell membrane, except that it is double layered. The nuclear envelope may have nuclear pores, which act as avenues of communication between the inner nucleoplasm and the outer cytoplasm. The nucleolus is a prominent, rounded nuclear organelle, which is usually centrally placed in the nucleoplasm when the cell is viewed microscopically. **(Bath-Balogh et al., ١٩٩٧).**

The cell organells have been described to include ~~**(Bath-Balogh et al. ١٩٩٧).**~~

Field Code Changed

~~T~~the endoplasmic reticulum (ER), which is parallel membrane-bound cavities in the cytoplasm that contain newly acquired and synthesized protein. Two types of ER, smooth surfaced and granular or rough surfaced, can be found in the same cell. Rough-surfaced ER is caused by ribosomes on the surface of the reticulum and is the site at which protein production is initiated. These ~~Ribosomes~~ ribosomes are particles that translate genetic codes for proteins and activate mechanisms for their

production. They can be found as separate particles in the cytoplasm, clustered, or attached to the ER membranes ~~.~~ **(Avery and Steele, ٢٠٠٠).**

~~**(Avery and Steele ٢٠٠٠)**~~

In addition, ~~The~~ the ER transport~~s~~ substances in the cytoplasm~~. The~~ ~~ER~~ and it is connected to Golgi's apparatus via small vesicles. Golgi's apparatus or complex helps sort, condense, package, and deliver proteins arriving from the ER. ~~Golgi's~~ Such apparatus is composed of cisternae (flat plates) or saccules, small vesicles, and large vacuoles. ~~From here the~~The secretory vesicles move or flow to the cell surface, where they fuse with the cell membrane and the plasmalemma and release their contents by exocytosis~~.~~ **(Avery and Steele, ٢٠٠٠).**

~~**(Avery and Steele ٢٠٠٠)**~~

Moreover ~~Lysosomes~~ lysosomes are small, membrane-bound bodies that contain a variety of acid hydrolase and digestive enzymes to help break down substances both inside and outside the cell~~.~~ **(Avery and Steele, ٢٠٠٠).**

~~**(Avery and Steele ٢٠٠٠)**~~

Furthermore, ~~Mitochondria~~ mitochondria are membrane-bound organelles that lie free in the cytoplasm. They are important in generating energy. These organelles appear as spheres, rods, ovoids, or thread-like bodies. Usually the inner layer of their trilaminar bounding membrane inflects to form transverse-appearing plates, the cristae~~.~~ **(Avery and Steele, ٢٠٠٠).**

~~**(Avery and Steele ٢٠٠٠)**~~

Surrounding the cell is the plasma membrane or plasmalemma, which envelops the cell and provides a selective barrier that regulates transport of substances into and out of the cell~~.~~ **(Avery and Steele, ٢٠٠٠).**

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