GENETIC DISORDERS OF THE PITUITARY GLAND

ESSAY

Submitted For Partial Fulfulment of

Master Degree In Pediatrics

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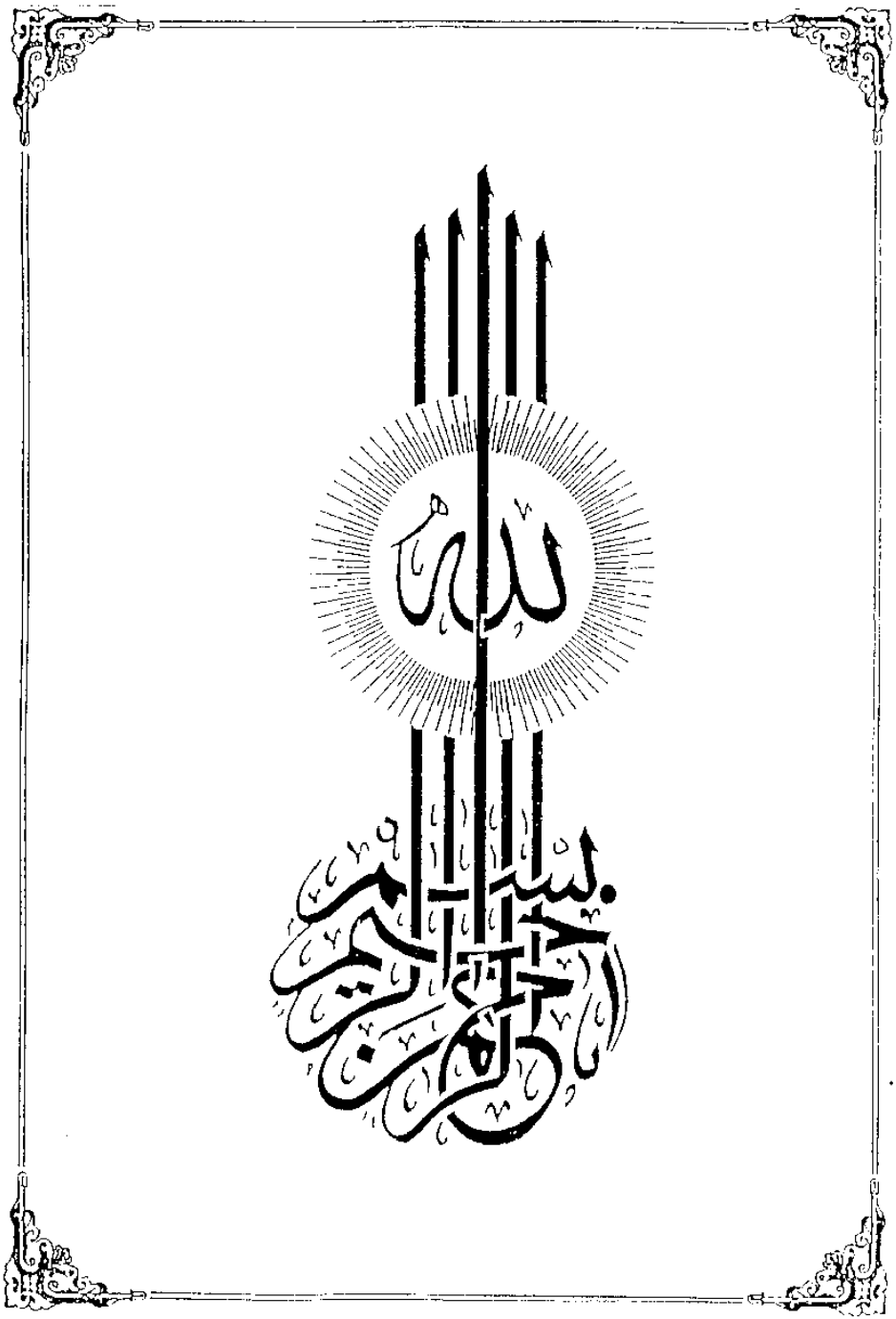
1990

ACKNOWLEDGEMENT

I would like first to express my deepest thanks and gratitude to Almighty GOD for his great help all the time.

I am grateful and indebted to my Chief Supervisor; Professor Dr. MOHAMED AHMED AWAD ALLA; Professor of Pediatrics, Faculty of Medicine, Ain Shams University, who suggested the subject of the Essay and also for his continuous help and encouragement during the performance of the work.

My very special and sincere thanks go to Dr. MOHAMED ABD EL-ADL EL-SAWI, Lecturer of Pediatrics (Genetics), Faculty of Medicine, Ain Shams University, who spared no time or effort in reading and criticizing, providing me with his long experience and discussing every detail in the Essay. His real encouragement has indeed given me a very good push. He was also too kind to provide me with a lot of references and books that opened new scopes before me.



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INTRODUCTION

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Vesalius first used the term pituitary (Latin: pituitslime) because of the ancient belief that effluent from brain was filtered through into the nasopharynx.

Soemmering described it as the hypophysis because of its location beneath the brain (John and Gavin, 1984).

The pituitary gland consists of an anterior lobe or adenohypophysis which includes the pars tuberalis and pars distalis, a vestigial middle lobe or pars intermedia and a posterior lobe or neurohypophysis which includes the median eminence, infundibulum and pars nervosa (John and Gavin, 1984).

Similar to the genetic disorders of the other endocrine glands, the genetic disorders of the pituitary gland involving the hormonal deficiency states are more common than hereditary forms of hyperpituitarism.

Pituitary deficiency disorders (hypopituitarism) may involve a single trophic hormone (monotrophic deficiency) or a combination of two or more pituitary hormones (multitrophic hormone deficiency) and may result from disturbances in any part of the hypothalamic hypophyseal-target organ complex (Rimoin and Horton, 1978 a, b).

Theoretically, a syndrome of pituitary hormonal insufficiency might result from; developmental, degenerative or receptor lesions of the hypothalamus, deficiencies of the hypothalamic releasing hormones or their receptors, developmental or degenerative lesions of the pituitary gland, deficiencies or structural abnormalities of the pituitary hormones, or defects in target organ responsiveness to hormonal action. Each of these mechanisms has now been described in patients with pituitary insufficiency, resulting in the marked genetic heterogeneity that has been observed in pituitary dwarfism (Rimoin, 1983).

Pituitary deficiency has been recognized as a cause of proportionate short stature and it is now apparent that pituitary dwarfism represents a heterogenous group of disorders secondary to a variety of genetic and acquired defects in human growth hormone (hGH) secretion or action (Rimoin, 1976; Rimoin and Schimke, 1971).

Also, isolated deficiencies, TSH, ACTH, LH, FSH and prolactin have been reported.

Hereditary vasopressin sensitive diabetes insipidus is a syndrome characterized by polyuria, polydipsia and dehydration secondary to a deficiency of antidiuretic hormone (Crawford and Bode, 1975).

Genetic disorders of pituitary hyperfunction are less common than those of pituitary insufficiency. Furthermore, except in instances of multiple endocrine adenomatosis or in patients who have a positive family history of pituitary hyperfunction, it is possible to denote which of the sporadic cases have a genetic form of pituitary disease.

The most common form of hereditary pituitary neoplasia is the multiple endocrine adenomatosis syndrome, Type I. Although multiple cases of familial acromegaly and of the amenorrhoea galactorrhoea syndrome have been described in certain kindreds with no evidence of other endocrine involvement, these disorders may well represent limited forms of the multiple endocrine adenomatosis syndrome (Rimoin, 1983).

Hypersecretion of vasopressin occurs in two different ways: in response to a recognizable non osmotic stimulus like hypotension and it is termed appropriate, the second type shows an abnormality in the osmoregulation of the hormone and it is termed inappropriate secretion (Syndrome of inappropriate secretion of ADH "SIADH") (Robertson et al., 1976).

It can be defined as incomplete suppression of the hormone in face of hypoosmolality of the body fluid (Robertson, 1981).

ANATOMY AND EMBRYOLOGY OF THE PITUITARY GLAND

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The pituitary gland is a small structure about 1 cm long; it weighs 500 mg and is divided into two parts; anterior (Adenohypophysis) and posterior (Neurohypophysis) (Jubiz, 1979). The adenohypophysis develops from ectodermal outgrowth of the stomadeum (primitive Buccal Cavity) known as Rathke pouch and is controlled by hypothalamic secretions. The neurohypophysis is derived from the infundibulum of the diencephalon from the floor of the third ventricle and it has direct neural connections via a large tract of fibres with neurons in the supra optic and para ventricular nuclei of the anterior hypothalamus (Behrman and Vaughan, 1987).

The pituitary lies in the sella turcica, a hollow of the sphenoid bone within a layer of dura mater named the diaphragma sella turcicae. Because this envelopment takes place early in fetal life, there are neither subarachnoid nor subdural spaces within the sella. The pituitary stalk is surrounded by subarachnoid space containing cerebrospinal fluid. On each side of the pituitary is the cavernous sinus. Anteriorly and posteriorly are small venous radicles cincturing the infundibulum. Anteriorly and just above the diaphragm lies the optic chiasma. Due to this relationship, disturbance of vision is frequently associated with tumours of the pituitary (John and Gavin, 1984).