Biliary Sludge and Stones in Mentally Retarded Patients.

THESIS

Submitted for partial fulfillment of
Master degree in Pediatrics

By

Abdel Rahman Ramadan Abdel Rahman
M.B.B.CH

Supervised by

Dr: Amani Osman Mahmoud
Professor of Pediatrics
Faculty of Medicine – Ain shams University

Dr: Osama Kamal Zaki
Consultant of Medical Genetics
Genetics Unit – Faculty of Medicine
Ain shams University

Faculty of Medicine
Ain shams University
2013

TO MY FATHER AND MOTHER

TO MY DEAR WIFE

TO MY LOVELY OMAR & JOOD

THIS WORK IS DEDICATED

abdelrhman

Acknowledgment

First and foremost, thanks ALLAH for whom I always pray to give me the ability to do my work faithfully.

I would like to express my sincere gratitude and deep appreciation to Dr. : Amani Osman Mahmoud, Professor of pediatrics, Faculty of medicine, Ain shams university for her helpful guidance, valuable suggestions, great patience and follow up throughout the research.

I am gratefully thankful to Dr. : Osama Kamal Zaki, Consultant of medical genetics, genetic unit, Faculty of medicine, Ain shams university for his generous advice, kind supervision, continuous support and helpful guidance during the conduction of this work

Many thanks to all members of the pediatrics department for their help and advice.

Many thanks to the laboratory and ultrasonography members participated in this work.

Thanks to all patients and their parents.

LIST OF ABBREVATIONS

AAI	Atlantoaxial instability
AAP	American Academy of Pediatrics
ABC-1	Aberrant Behavior Checklist
ACOG	American College of Obstetricians and Gynecologists
AFP	Alpha -Fetoprotein
AML	Acute myeloid leukemia
BEAR	Brainstem auditory evoked response
CBD	Common bile duct
CNS	Central nervous system
CVS	Chroionic villus sampling
DSM-1V-TR	The Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision

ERS	Endoscopic retrograde sphincterotomy
GI	Gastrointestinal
hcG	Human Chorionic Gonadotropin
ID	Intellectual disabilities
IQ	Intelligence quiescent
MR	Mental retardation
MRI	Magnetic resonanance imaging
NH	Neonatal hemochromatosis
NHANES	The National Health and Nutrition Examination Survey
NIH	National institute of health
NT	Nuchal Transluceny
OAE	Otoacoustic emission
PKU	Phenylketonurea
RUQ	Right upper quadrant
TDM	Transient myeloproliferative disorder
TL	Transient leukemia
TSH	Thyroid stimulating hormone
UCDA	Ursodeoxycholic acid
US	United states

CONTENTS

LIST OF TABLES

LIST OF FIGURES

INTRODUCTION

Mental retardation (MR) refers to a group of disorders that have in common deficits of adaptive and intellectual function and an age of onset before maturity is reached. **(Bruce et al., 2004).**

IQ scores of Down's syndrome patients range from 25 to 75 with an average of 40 to 45 in young adults. **(Peter, 2005).**Before the 1980s, few cases of cholelithiasis in children, relating to hemolytic anemia, had been described in the medical literature. The use of ultrasonography increased the frequency of diagnosis of cholelithiasis in childhood .The prevalence of cholecystolithiasis may be increasing in childhood and adolescence **(Kang et al., 2003).**

An overall prevalence of 0.13% to 1.9% for gallstone disease in children up to 19 years of age was reported **(Herzog and Bouchard , 2008).**

Some reports have showed that biliary abnormalities like lithiasis and biliary sludge occur more frequently among Down syndrome patients. **(Toscano et al.,2001)**

AIM OF THE WORK

The aim of this study is to detect the prevalence of biliary sludge and stones among mentally retarded patients.

GALLSTONE

Background

Gallbladder disease is one of the most common and costly digestive diseases that requires hospitalization in the United States. Gallbladder calculi are more common in the adult population and remain relatively uncommon in children; however, the incidence of cholelithiasis in children has increased. **(George and David , 2009).**

There are over 700,000 cholecystectomies (removal of the gallbladder) performed in the United States each year, Complications are not common and mortality from the disease is very, very low. There are risk factors that increase the chances of developing gallstones– e.g., gender, age, and genetic traits – that cannot be changed, but there are many lifestyle risk factors, as well. **(Shaffer, 2006)**

Gallstone formation is therefore very poorly understood. Surprisingly, in the last few decades there

has been significant rise in gallstone disease among children. **(Kumar et al., 2000).**

Choledocholithiasis refers to the presence of gallstones within the common bile duct (CBD). **(Everhart et al., 1999).**

Sludge is defined as an echogenic material within the gallbladder that layers and changes shape and position on moving the patients. **(Winter et al., 1994)**

Normal common bile duct diameter between 5-8mm. Allowance for a larger diameter is sometimes accepted in older patients**. (Daradkeh et al., 2005)**

The diameter of the duct may increase or decrease following cholecystectomy, but the change is generally not significant. **(Channa et al., 2007 & Kim et al., 2003)**

Characteristics and composition

The formation of gallstone in vivo takes years and it is quite difficult to monitor such events from nucleation to the consolidation. **(Swidsinski et al., 1998)**

Gallstones can vary in size and shape from as small as a grain of sand to as large as a golf ball. **(George & David, 2009)**

On the basis of their composition, gallstones can be divided into the following types:

- **Cholesterol stones** are formed from cholesterol supersaturation of bile and are composed of 70-100% cholesterol with an admixture of protein, bilirubin, and carbonate. These account for most gallstones in adults but make up only about 21% of stones in children. **(National Institute of Diabetes and Digestive and Kidney Diseases , 2007).**

Cholesterol stones vary in color from light-yellow to dark-green or brown and are oval 2 to 3 cm in length, often having a tiny dark central spot. To be classified as such, they must be at least 80% cholesterol by weight (or 70%, according to the Japanese classification system **(Kim et al., 2003)**

There are three reasons for the formation of cholesterol gallstones:

Cholesterol supersaturation of bile:

Bile acids and phospholipids normally help keep cholesterol soluble so that it does precipitate out. However, there are times when the ratio of bile acid and phospholipids to cholesterol in the bile is altered, the bile becomes supersaturated with cholesterol, and it crystallizes into cholesterol gallstones. This can happen when there is hypersecretion of cholesterol into bile by the liver (the most common cause) or hyposecretion of bile salts or phospholipids.