*I am deeply thankful to "**Allah**" by the grace of whom, this work was possible.*

*I would like to express my deepest appreciation and gratitude to **Prof. Dr. Sahar Mohammed Elfekky,** Professor of Radiodiagnosis, Ain Shams University, for her kind supervision, valuable advises and constructive guidance were the real driving force in the initiation progress and completion of this thesis.*

*I am greatly indebted to **Dr. Mennatallah Hatem Shalaby,** Lecturer of Radiodiagnosis, Ain Shams University, for her greatest and unforgettable help and for her valuable comments, suggestions and close supervision. She dedicated much of his effort and time for the direction of this work.*

*My profound thanks go to **Ass. Prof. Rasha Tarif Hamza,** Assistant Professor of Pediatrics, Ain Shams University, for her indispensable help and continuous encouragement. Her effort was the backbone of the practical part of this work.*

Lastly but not the least I should like to express my appreciations to the patients for their cooperation throughout my work.

Dunya BurhanAldin Abdulla


وأنزل الله عليك
الكتاب والحكمة
وعلَّمَكَ مَا لَمْ تَكُن
تَعْلَمُ وكَانَ فَضْلُ
اللَّهِ عَلَيْكَ عَظِيماً
صدق الله العظيم
سورة النساء الآية
(١١٣)

RENAL ANOMALIES IN PATIENTS WITH TURNER SYNDROME: ROLE OF SCINTIGRAPHY

Thesis

Submitted for partial fulfillment of M.Sc. Degree in Radiodiagnosis

By

Dunya BurhanAldin Abdulla

M.B.B.Ch.

Supervised By

Prof. Dr. Sahar Mohammed Elfekky

Professor of Radiodiagnosis

Faculty of Medicine - Ain Shams University

Dr. Mennatallah Hatem Shalaby

Lecturer of Radiodiagnosis

Faculty of Medicine - Ain Shams University

Ass. Prof. Rasha Tarif Hamza

Assistant Professor of Pediatrics

Faculty of Medicine - Ain Shams University

Department of Radiodiagnosis
Faculty of Medicine
Ain Shams University

2014

تشوهات الكلى في مرضى متلازمة تيرنر:دور المسح الذري

توطئ للحصول على درجة الماجستير في
الاشعة التشخيصية

دراسة مقدمة من الطبيبة

دنيا برهان الدين عبدالله

بكالوريوس طب وجراحة عامة

تحت اشراف

أ.د. سحـــر محمـــد الفقـــي

استاذ الاشعة التشخيصية

كلية الطب – جامعة عين شمس

د.منـــة الله حاتـــم شلبـــي

مدرس الاشعة التشخيصية

كلية الطب – جامعة عين شمس

ا.م.د.رشـــا طريـــف حمـــزة

استاذ مساعد طب الاطفال

كلية الطب–جامعة عين شمس

قسم الاشعة التشخيصية
كلية الطب
جامعة عين شمس

2014

List of Contents

List of Abbreviations

3D ..... Three-dimensional

ACE ..... Angiotensin converting enzyme

ACR..... American college of radiology

CTU ..... Computed tomography urography

DMSA ..... Dimercaptosuccinic acid

DTPA ..... Diethylene triamine penta-acetic acid

ERPF ..... Effective renal plasma flow

FISH ..... Fluorescence in situ hybridization

GFR ..... Glomerular filtration rate

LEAP/GAP ..... Low-energy all-purpose/general all-purpose

MR ..... Magnetic resonance

OIH ..... Orthoiodohippurate

OM ..... Otitis media

SENSE ..... Sensitivity encoding

SHOX ..... Short-stature homeobox-containing gene

SPAIR q..... Spectral presaturation with inversion recovery

SPECT ..... Single-photonemission computed tomography

SPGR ..... Spoiled gradient echo

SSFS ..... Single-shot fast spin echo

STIR ..... Short-tau inversion recovery

TS ..... Turner syndrome

List of Tables

List of Figures

INTRODUCTION

Turner syndrome (TS) is diagnosed in females with partial or complete absence of one X chromosome (45, X karyotype). Clinical manifestations vary and may be subtle, but they usually include short stature, a broad chest with widely spaced nipples, cubitus valgus, congenital lymphedema, and a lack of spontaneous pubertal development from ovarian sex hormone insufficiency ***(Jones and Smith, 2006).***

Turner syndrome occurs in one out of 2,500 to 3,000 live female births; however, many more 45X conceptuses do not survive past the first trimester. Ninety-nine percent of conceptuses with a 45X karyotype abort spontaneously; Turner syndrome causes 10 percent of all first-trimester miscarriages ***(Menasha et al., 2005)***

Unlike with Down syndrome, maternal age does not increase the risk of Turner syndrome, and there are no clearly established risk factors. Recurrence in subsequent pregnancies is rare ***(Gardner and Sutherland, 2004).***

The presentation of Turner syndrome varies throughout a patient's life. The diagnosis should be considered in a female fetus with hydrops, increased nuchal translucency, cystic hygroma, or lymphedema ***(Sanders and Blackmon, 2002).***

A diagnosis of Turner syndrome can be confirmed with standard karyotyping (i.e., chromosomal analysis of 30 peripheral lymphocytes). More than one half of patients with the condition will have a missing X chromosome (45, X) in all cells studied or a combination of monosomy X and normal cells (45, X/46, XX; mosaic Turner syndrome) ***(Bondy, 2007).***

A mosaic result does not necessarily predict severity because karyotyping only investigates lymphocytes, not the relevant tissues (e.g., brain, heart, ovaries). ***(Bondy, 2007).***

Congenital malformations of the urinary system are present in up to 30% of patients with TS. Rotational abnormalities and double collecting systems are found most frequently. Although many of these abnormalities do not have clinical significance, some may result in an increased risk of hypertension, urinary tract infections, or hydronephrosis***(Lippe, 2009).***

Therefore, all individuals with TS should have a renal ultrasound study performed at the timeof diagnosis. If abnormalities are detected, further evaluations should be performed, and the appropriate therapy instituted. Additionally, in such individuals, ultrasound and urine cultures should be performed every 3–5 years ***(Lippe, 2009).***

Radionuclide renography refers to serial imaging after intravenous administration of technetium-99m DTPA or technetium-99m MAG3. It is used for qualitative and quantitative

evaluation of differential renal function. A commonly used technique involves dynamic acquisition of 1-2 second images for 1 minute (vascular phase), followed by 15-60 second images for 20 to 30 minutes (functional uptake, cortical transit, and excretion phases).If evaluation of renal perfusion is not needed, the examination is performed without the first phase ***(Boubaker et al., 2006).***

The regions of interest are typically drawn around the whole kidneys, but occasionally are limited to the renal cortex if a considerable amount of collecting system activity is present. A background region of interest is placed adjacent to each kidney***(Boubaker et al, 2006).***

Depending on the regions of interest drawn, the time-activity curves will reflect the functional clearance of the whole kidney, renal cortex, or collecting system. The differential renal function is calculated based on the relative counts accumulated in each kidney during the second minute after injection of the radiopharmaceutical ***(Boubaker et al, 2006).***

AIM OF THE WORK

- To assess the frequency of renal anomalies detected by scintigraphy in patients with Turner syndrome.
- To assess the advantages of renal scintigraphy over abdominal ultrasound and its role in early detection of renal affection in patients of Turner syndrome.
- To evaluate split renal function in such patients.

NORMAL RENAL ANATOMY AND RELEVANT RENAL ANOMALIES

A- Outline anatomy

The kidneys are retroperitoneal organs that are located in the perirenal retroperitoneal space with a longitudinal diameter of 10–12 cm and a latero-lateral diameter of 3–5 cm and a weight of 250–270 g. The kidney initially develops opposite to the future S2 vertebra, but eventually comes to rest opposite the L1 or L2 vertebra ***(Federle, 2006).***

In the supine position, the medial border of the normal kidney is much more anterior than the lateral border, so that the kidneys lie at an angle of about 30° from the horizontal ***(Nino-Murcia et al., 2000).***

The upper pole of each kidney is situated more posteriorly than the lower pole.

Renal anatomical Relations

I- anteriorly

The right kidney,, has a relation with the inferior surface of the liver with peritoneal interposition, and with the second portion of the duodenum without any peritoneal interposition since the second portion of the duodenum is retroperitoneal. **(Fig. 1a)** ***(Quaia et al., 2011).***

The left kidney has a relation with the pancreatic tail, the spleen, the stomach, the ligament of Treitz and small bowel, and with the left colic flexure and left colon **(Fig. 1a).** Over the left kidney, there are two important peritoneal reflections, one vertical corresponding to the spleno-renal ligament (connected to the gastro-diaphragmatic and gastrosplenic ligaments) and one horizontal corresponding to the transverse mesocolon ***(Quaia et al., 2011).***

II- Posteriorly

Both kidneys present a relationship with the diaphragm, with the lateral margin of the psoas muscle, with the aponeurosis of the transverse abdominis muscle, and with the lumbar muscle **(Fig. 1b).** ***(Quaia et al., 2011).***

III-Superiorly

Both kidneys have a relation with the adrenal glands, while the right kidney is separated from the inferior surface of the liver by the interposition of a double peritoneal sheet which derives from the reflection of the peritoneum to the inferior limit of the coronary liver ligament that delimitates the hepatorenal space or Morrison pouch ***(Quaia et al., 2011).***

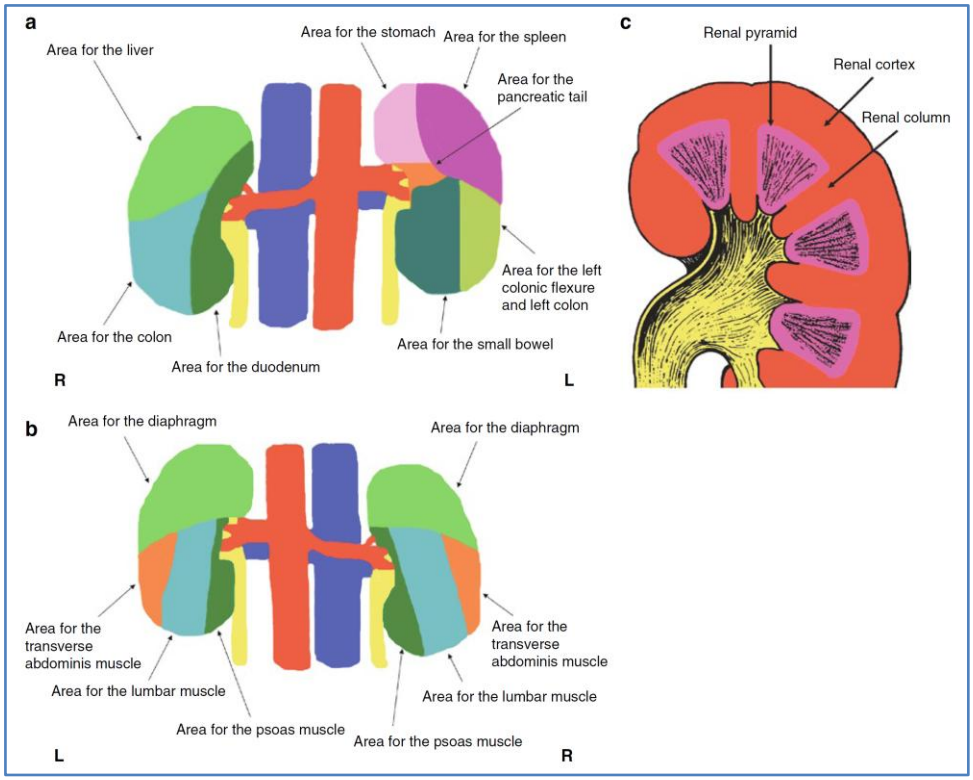

Fig. (1): (a) Scheme of the anterior anatomical relations of the kidneys: R right; L left. (b) Scheme of the posterior anatomical relations of the kidneys: R right; L left. (c) Scheme of the main components of the renal parenchyma (renal cortex, renal columns, and renal medulla divided in multiple renal pyramids) overlied by the renal capsule (black color) ***(quoted from Quaia et al., 2011).***

B- The renal parenchyma

The renal parenchyma is composed of different components. **(Fig. 1c).**

The renal capsule is a tough fibrous layer surrounding the kidney (fibrous renal capsule) and covered by a thick layer of perinephric adipose tissue (fat renal capsule). The kidney is covered by the renal capsule formed by the fibrous and adipose