معهد بيولوجيا بيطرية

Prof. Dr. Mohamed A. Saber
Ass. Prof. Dr. Ehab El-Dabaa
Dr. Mohamed Abbas Shemis

DEPARTMENT OF BIOCHEMISTRY
Biotechnology and Genetic Engineering Unit
Molecular Diagnosis Lab.

Name: ندى محمد محمد احمد **Code No:**

Referred by:

Sample:

Date: 10/4/2010

Cystic fibrosis by PCR
(Δ F 508 mutation)

	Reference Range
Result: Heterozygous mutation	**Negative**

Date:18/4/2010 **Signature**

Kornish EL-Nile. Warak EL-Hadar. 12411 Giza – P.O. BOX 30 Imbaba
Tel/Fax: (02) 35402977 Tel: 35407276-35401019

Cairo University
Specialized Pediatric Hospital
Pulmonary Function Unit

Name: ندى محمد محمد
Date: 31/12/2009
Age: 10years.
Sex: female
Sweat Chloride Test: 104.8 mEq/L

(Normal 0- 40 mEq /L)

(Borderline 40-60 mEq/L)

(Abnormal > 60 mEq/L)

Head of the department
Prof. Dr. Mona El Falaki

Done by:
Dr. Mona Mohsen

Kasr El Aini

Kasr Al Ainy Faculty of Medicine - Cairo University كلية طب القصر العينى- جامعة القاهرة

New Children's Japanese Hospital

Department of Clinical and Chemical Pathology - Molecular Biology unit

Patient Name: ندى محمد محمد احمد حسن Age: years

Referred by: Date:18 /12/2019

Genetic assay of the CFTR gene for the diagnosis of Cystic fibrosis

Test spectrum:

The test covers the following mutations using PCR and Reversed Hybridization

CFTRdel2 , 3	W1282X	G85E	1507del (-ATC)
2183AA>G	R334W	394delTT	IVS85T/7T/9T
2142delT	R560T	A455E	2184delA
R117H	F508del (-CTT)	2184insA	189+1G>A
Y122X	1717-1 G>A	2789+5G>A	3120+1G>A
621+1G>T	R1162X	3272-26A>G	711+1G>T
G551D	3659delC	Y1092X	1078del
3849+10kbC>T	R347H	R334W	189+1G>A
G542X	R553X	390insT	A455E
N1303K	3120+1G>A	3272-26A>G	Y1092X
3849+10kbC>T			

Comment:

The patient expresses a **Compound Heterozygous genotype for the mutations F508del (-CTT) and W1282X** in the CFTR gene.

Polymorphism in the IVS 8 of the CFTR gene is (7T/9T).

رئيس الوحدة

Professor Dr/ Manal Wilson

Alfa Pneumo Care

مركز ألفا للأمراض الصدرية

Medical Report

Name: Nada Mohamed Mohamed
Age: 19 years old
Diagnosis: Cystic Fibrosis, pancreatic insuffiency, bronchiectasis.

Ms Nada is 19 years old female patient, was diagnosed since early childhood as cystic fibrosis, with a well known family history of such disease.
She had bilateral advanced bronchiectasis, frequent pulmonary exacerbation and previous ICU admission with use of non-invasive ventilation previously.
She had also pancreatic insuffiency.
She needs the following medications for her condition as a long term lifelong therapy:

Dornase alpha (pulmozyme) 2.5 mg once daily
Tobramycin inhalation alternating month bid
Creon tablets 2-3 tab before heavy meals 1 tab before snacks

She needs regular follow up every three months in a specialized chest clinic.

Prof/ Yosri Akl
Professor of pulmonary medicine
Cairo University

Dr/ Mohamed Said
Lecturer of Pulmonary Medicine
Cairo University

مركز الفا للأمراض الصدرية
١ ميدان الحجاز - برج الصفا الطبى المهندسين
رقم بطاقة ضريبية: ٢٠٥-٠٢٢-٦٥٠
رقم ملف ضريبى: ٥-٠٠٠٨٥-٥٣١-٠٠٠
مأمورية ضرائب الإستثمار
سجل تجارى: ١١٩٢٤٢
تليفون:- ٣٣٣٨١٠٤٠ (٢٠٢) - ٣٣٣٨٣٥٣٥ (٢٠٢)

NILE
SCAN & LABS

مركز النيل للأشعة والتحاليل

Name : ندي محمد محمد

Date : 11/03/2021
Code No. : 11713841

ABDOMEN ULTRASONOGRAPHIC SCANNING REVEALED:

Liver is of average size. Parenchyma displays heterogeneous bright echopattern. No focal lesions or intrahepatic biliary dilatation are seen.
Portal vein patent and of average caliber
Gall bladder is of normal size. No evidence of calculi.
Spleen is mildly increase in size and showing homogenous echo pattern with no focal lesions. It measures 13.6 cm in its longitudinal axis.
Pancreas and para-aortic region are concealed by gases.
Both kidneys are of normal size and shape showing mild increase echopattern with preserved cortico-medullary differentiation . No back pressure changes. No stones or masses are seen.

Right kidney measures = 10.8x5.6 cm with parenchymal thickness = 16 mm.
Left kidney measures = 10x3.6 cm with parenchymal thickness = 11 mm.

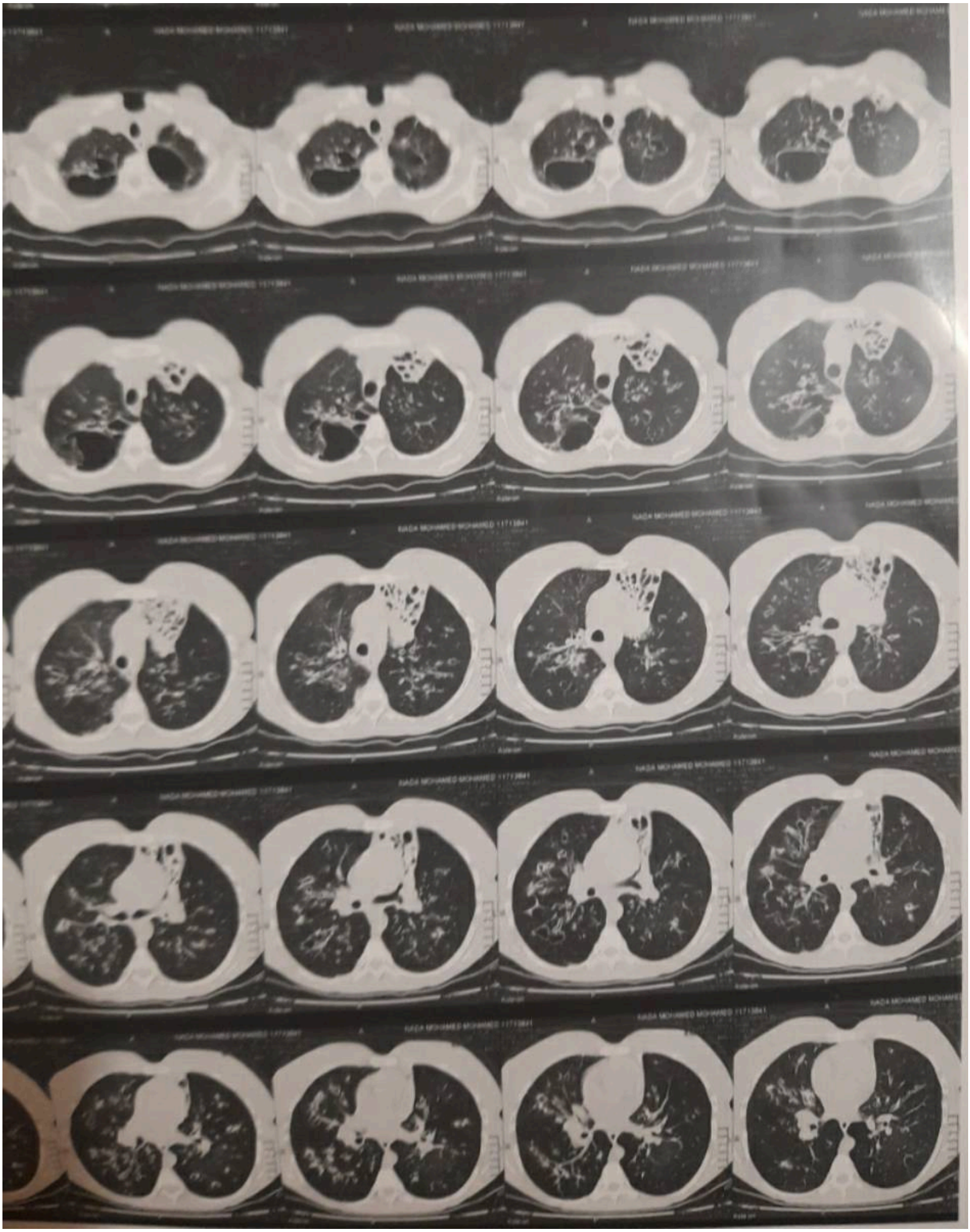
No ascites.

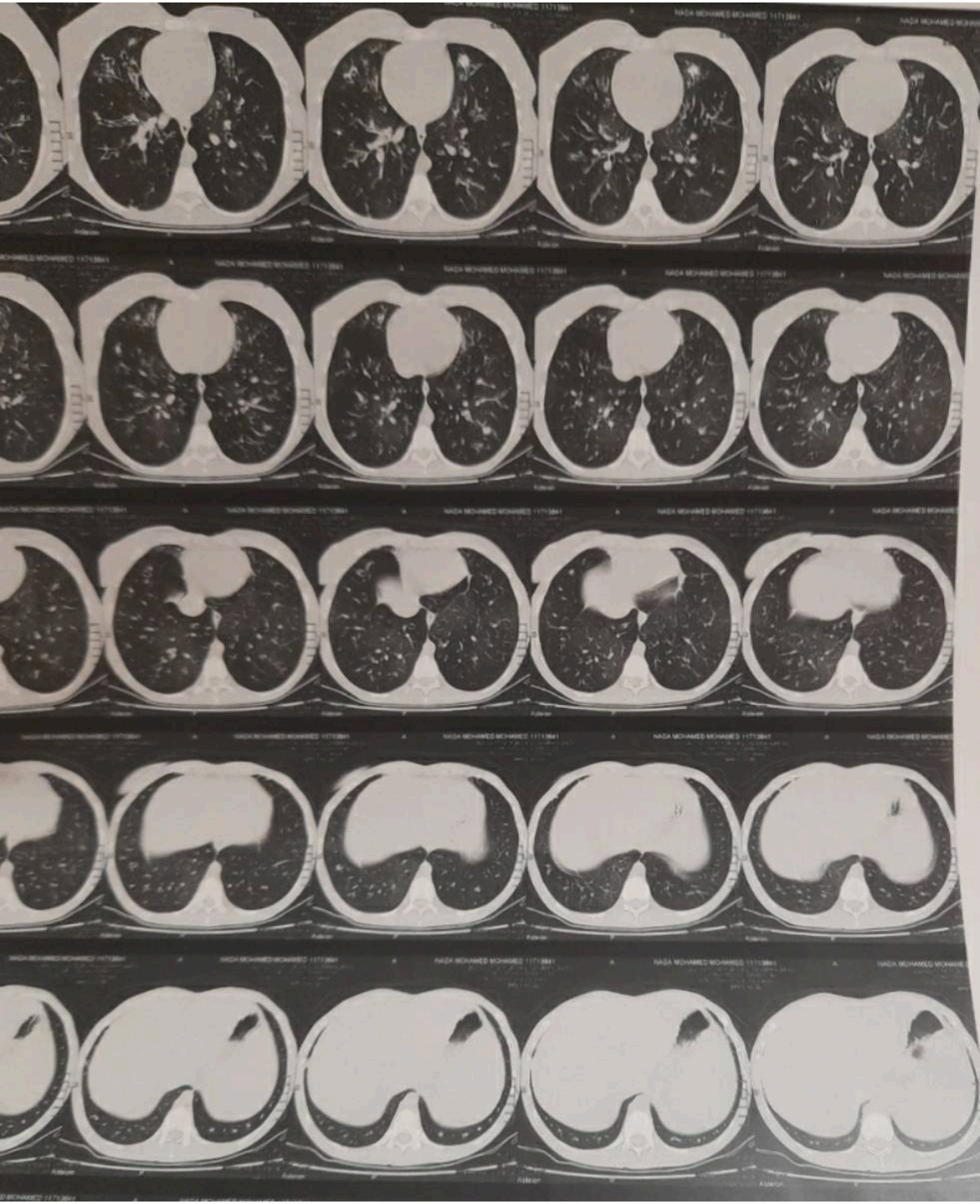
CONCLUSION:-

- ***bright heterogeneous hepatic parenchymal echopattern for clinical and laboratory correlation .***
- ***Bilateral parenchymatous renal disease for clinical and laboratory correlation.***
- ***Splenomegaly***

Dr.Sara Moustafa.MSc

NILE DIAGNOSTIC CENTER





NILE SCAN & LABS

مركز النيل للأشعة والتحاليل

DIAGNOSTIC CENTER

Name : ندي محمد محمد

Date : 11/03/2021

Code No. : 11713841

NON-CONTRAST HIGH RESOLUTION MDCT OF THE CHEST WITH CORONAL AND SAGITTAL REFORMATTING REVEALED:-

Clinical background: Known case of cystic fibrosis

Findings:

compared to the previous study dated 18-02-2020 the current study show

- stationary course as regards the previously noted hyper inflated chest with bilateral apical emphysematous bulla ,largest on the left side measuring 4.7xcm
- Rather stationary status of the bilateral pulmonary architectural distortion and mosaic appearance with the widely spread areas of bronchiectasis with predilection in upper, middle and apical segments of lower lobes.
- Left upper lobe anterior segment consolidation area with cystic air bronchogram seen within is still noted
- No hilar or mediastinal masses or lymph node enlargement.
- No pleural or pericardial collection, masses or calcification.
- Normal plain CT appearance of the heart and great vessels.
- Patent trachea and major tracheo-bronchial airways.
- Intact bony thoracic cage.
- Still noted cirrhotic liver.

OPINION:

Known case of cystic fibrosis, compared to the previous study dated 18-12-2020 the current study show:

- stationary course as regarding hyper inflated chest with apical segmental emphysematous bullae
- Rather stationary status of the Left upper lobe anterior segment consolidation area with cystic air bronchogram
- Stationary status of bilateral pulmonary architectural distortion and mosaic appearance with widely spread areas of bronchiectasis
- For clinical correlation , further assessment and close follow-up

A.Prof.Mohamed Hafez,MD&PhD