UNIQUE OCCURRENCE OF GORLIN-GOLTZ SYNDROME WITH SITUS INVERSUS TOTALIS

A DIAGNOSTIC ENIGMA

GORLIN GOLTZ SYNDROME

Firstly reported by JARISH and WHITE IN 1894
Incidence :1:57000

inherited as an autosomal dominant trait with variable expressivity

FIRST TIME REPORTED IN WORLD LITERATURE BY DR. MONIKA GUPTA

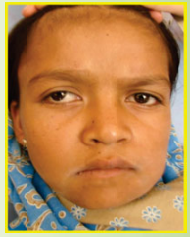
Coincidental occurrence is unlikely

SITUS INVERSUS TOTALIS

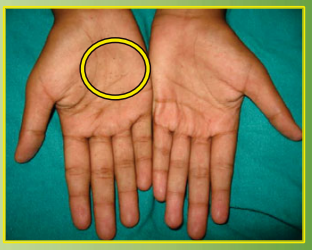
Means complete mirror image of thoracic and abdominal organs

Firstly described by BAILLIE in 18th century
Incidence 1: 10000

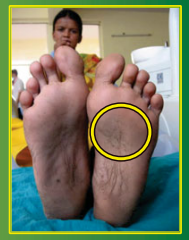
CLINICAL FEATURES



Facial Asymmetry , Frontal bossing and Hypertelorism



Palmer Keratosis



Planter Keratosis

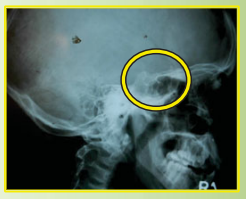
Clinical phenotypes are histogenetically linked to Patched Tumor Suppressor Gene (PTCH) / Hedgehog Signalling

PTCH localized to primary cilia and mediate key steps in transduction of the Hedge hog signal

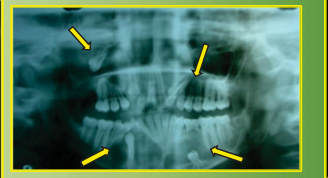
INFERENCE

Ciliary Dysfunction via dysregulation of the Hedgehog pathway is the underlying cause of both Situs Inversus Totalis and Gorlin-Goltz

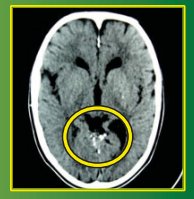
RADIOLOGICAL FEATURES



Lateral view of skull showing BRIDGING of the SELLA TURCICA

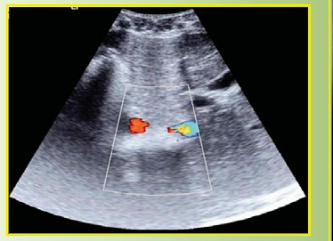


OPG showing multiple EXPANSILE CYSTIC LESIONS in the maxilla and mandible

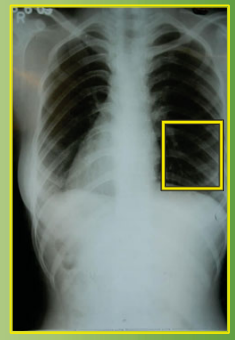


Axial CT Scan showing CALCIFICATION of FALX CEREBRI ALONG VENOUS SINUSES

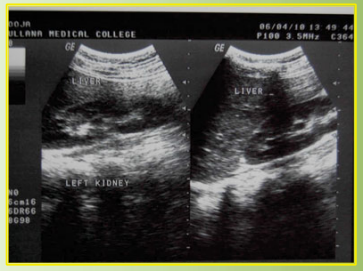
RADIOLOGICAL FEATURES



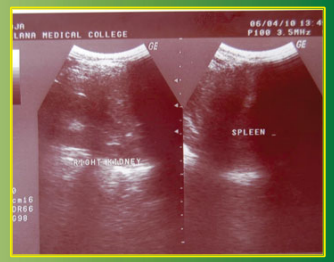
USG showing Transposition of Great Vessels



P A View of Chest showing Bifid Fourth and Fifth Ribs on the left side & Dextro-cardia



USG showing Liver in the Left Hypochondrium



USG showing Spleen in the Right Hypochondrium

PRESENTED BY:
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