Fig A

DIAGNOSTIC CRITERIA FOR NEUROFIBROMATOSIS TYPE I
CLINIC DIAGNOSIS

5 OR MORE CAFE'-AU-LAIT-SPOTS (>0,5 cm BEFORE PUBERTY, 1,5 cm AFTER PUBERTY)

2 CUTANEUOS/SUB-CUTANEUOS NEUROFIBROMAS OR PLEXIFORM NEUROFIBROMA

AXILLARY AND INGUINAL FRECKLING

AN OPTICAL GLIOMA

2 OR MORE LISCH NODULES

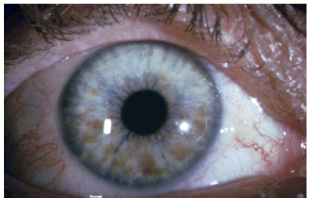
ORTOPHEDIC ABNORMALITIES (SPHENOID WING DYSPLASIA, TIBIAL PSEUDOARTHROSIS, SCOLIOSIS)

FIRST DEGREE RELATIVE WITH NF1

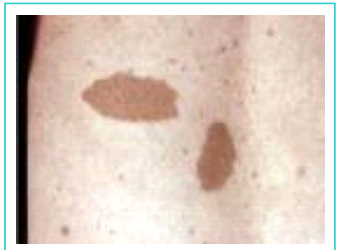
NATIONAL INSTITUTE OF HEALTH CONFERENCE 1988

Fig B

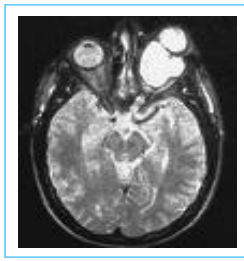
CLINICAL MANIFESTATIONS OF NEUROFIBROMATOSIS TYPE I



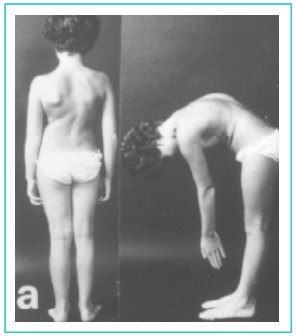
LISCH NODULES



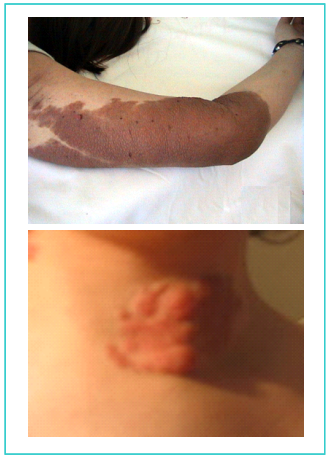
CAFE'-AU-LAIT-SPOTS



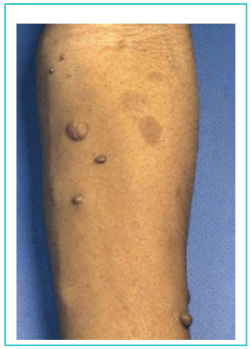
OPTICAL GLIOMA



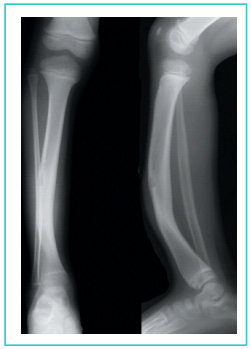
SCOLIOSIS



PLEXIFORM NEUROFIBROMA



CUTANEOUS NEURPFIBROMAS



OSSEOUS LESIONS

Fig C

NF1 GENE STRUCTURE AND LOCALIZATION

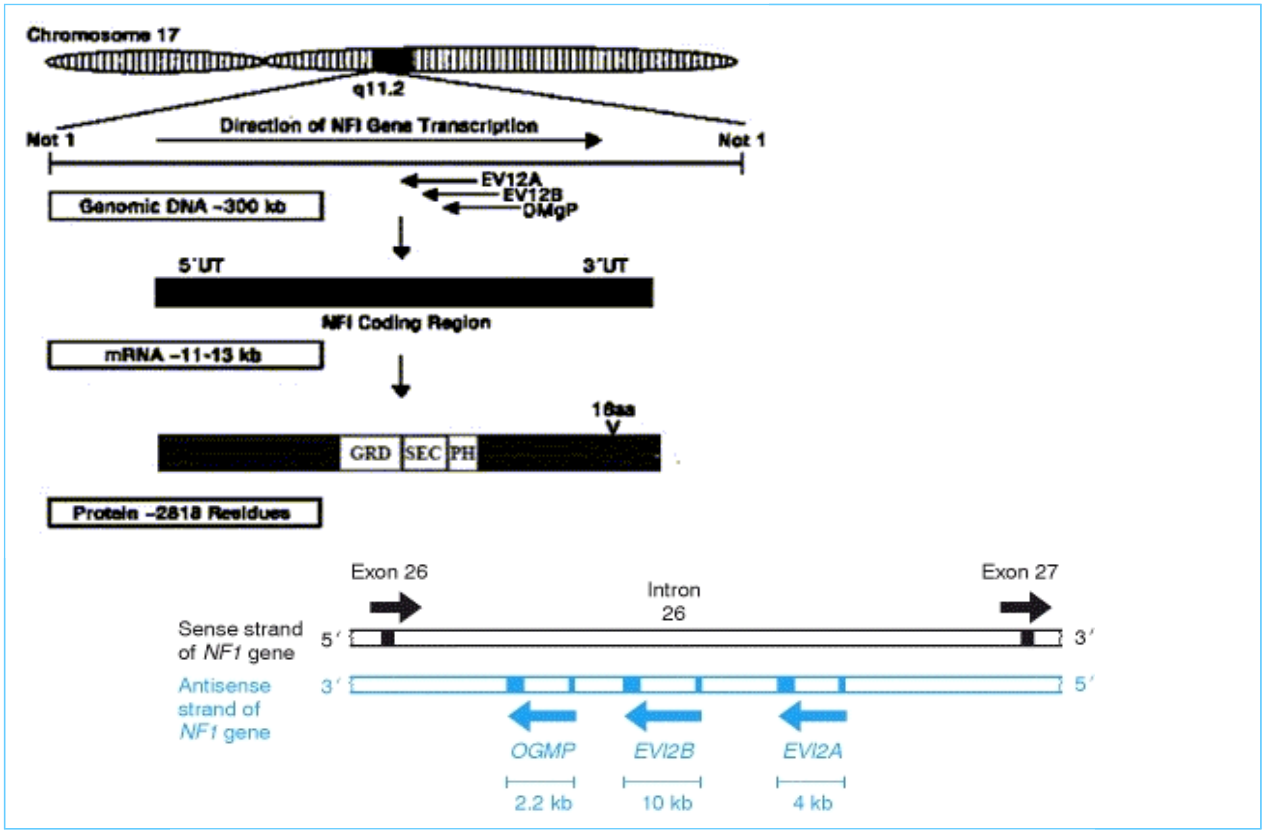


Fig D

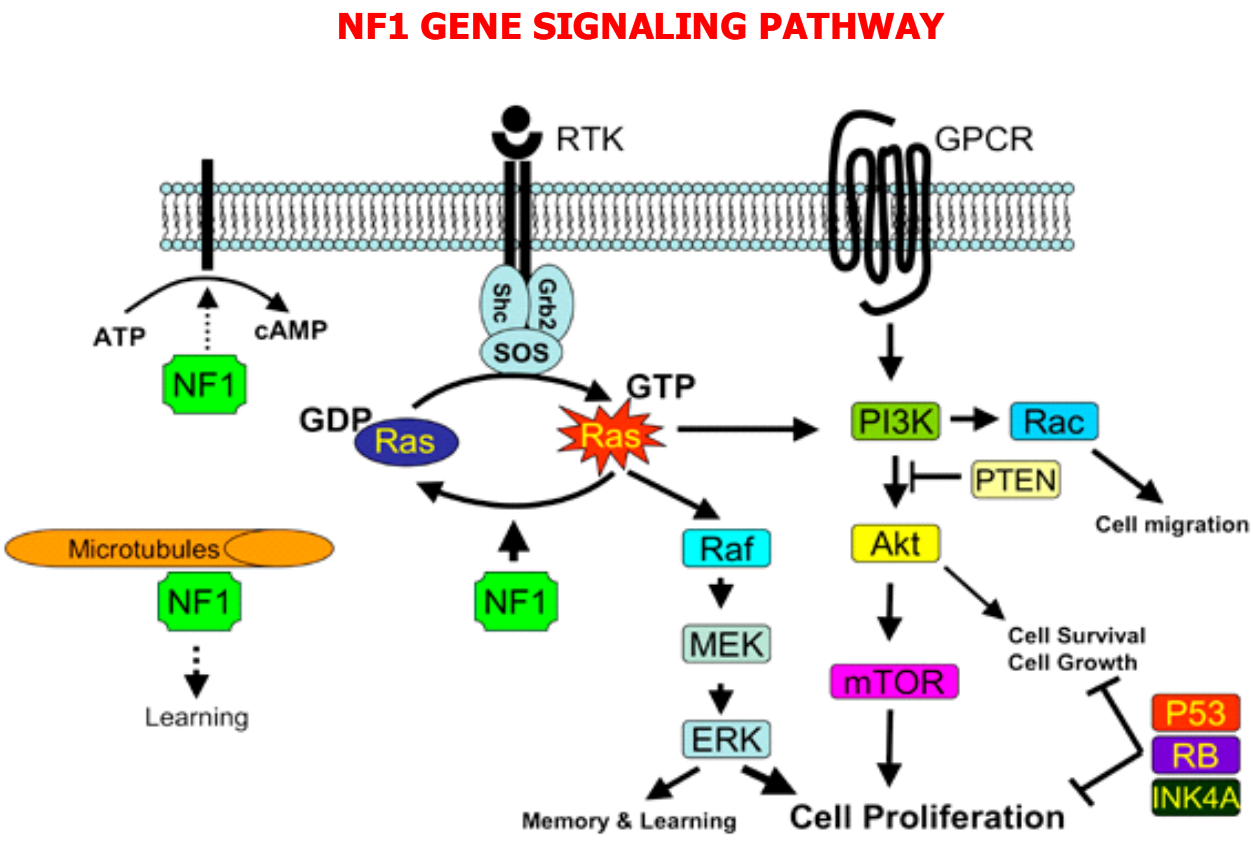
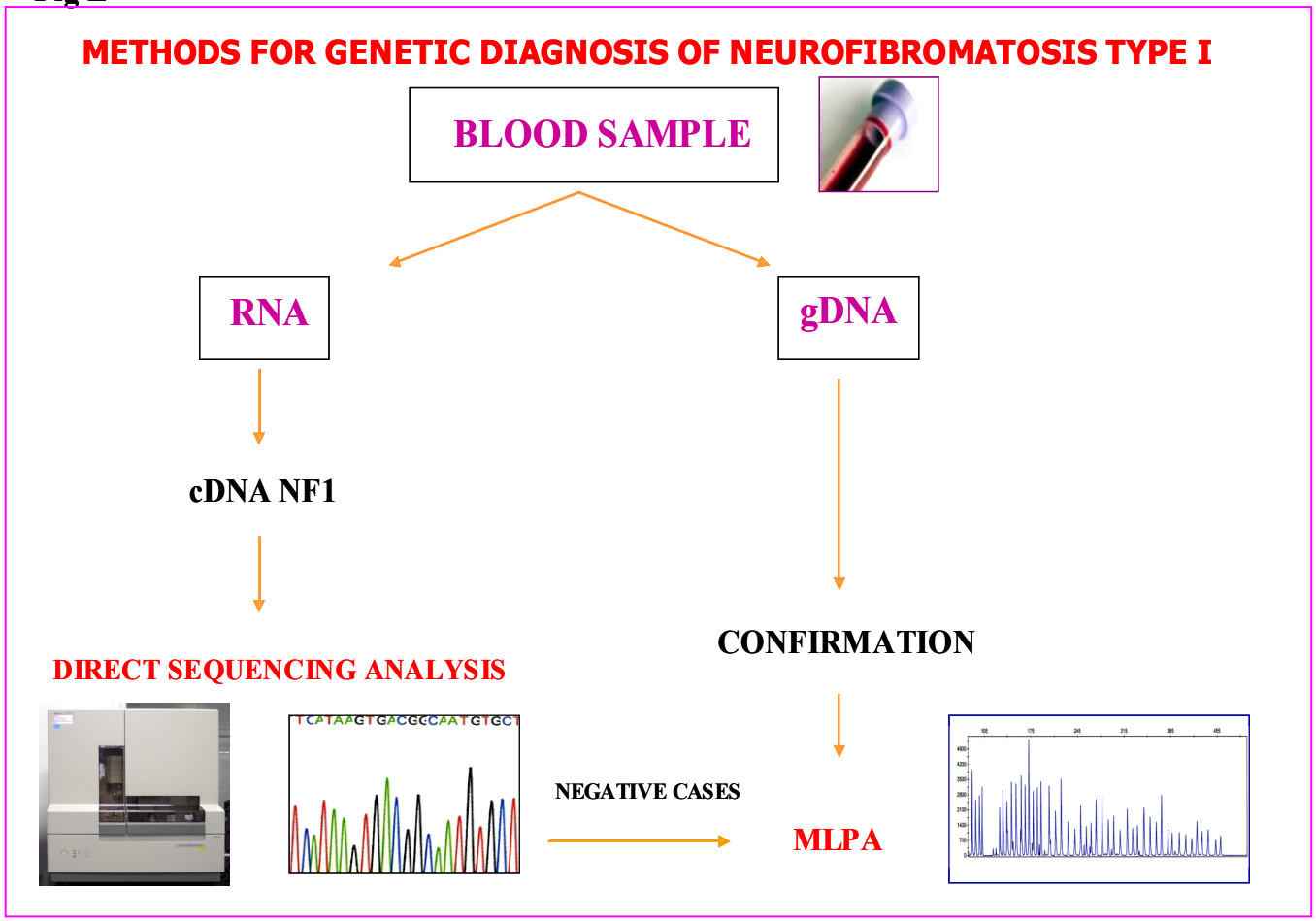

NF1 GENE SIGNALING PATHWAY
RTK
GPCR
Shc
Grb2
SOS
ATP
cAMP
NF1
GDP
Ras
GTP
Ras
PI3K
Rac
PTEN
Cell migration
Microtubules
NF1
Learning
NF1
Raf
Akt
MEK
Cell Survival
Cell Growth
mTOR
P53
RB
INK4A
ERK
Memory & Learning
Cell Proliferation

Fig E


METHODS FOR GENETIC DIAGNOSIS OF NEUROFIBROMATOSIS TYPE I
BLOOD SAMPLE
RNA
gDNA
cDNA NF1
CONFIRMATION
DIRECT SEQUENCING ANALYSIS
NEGATIVE CASES
MLPA