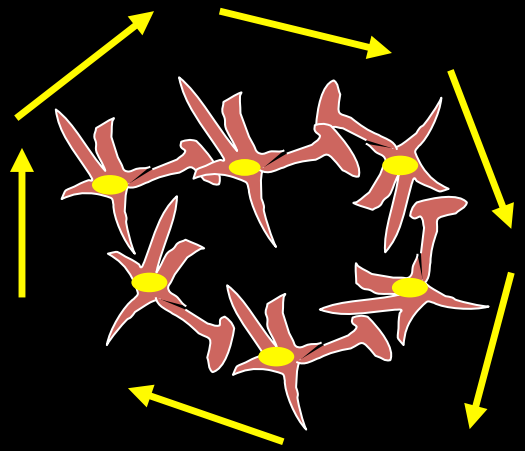
U.O.P.I. di Malattie Rare del Sistema Nervoso in Età Pediatrica
Scuola di Specializzazione in **Pediatria**
Dipartimento di MEDICINA CLINICA e SPERIMENTALE
Università degli Studi di Catania

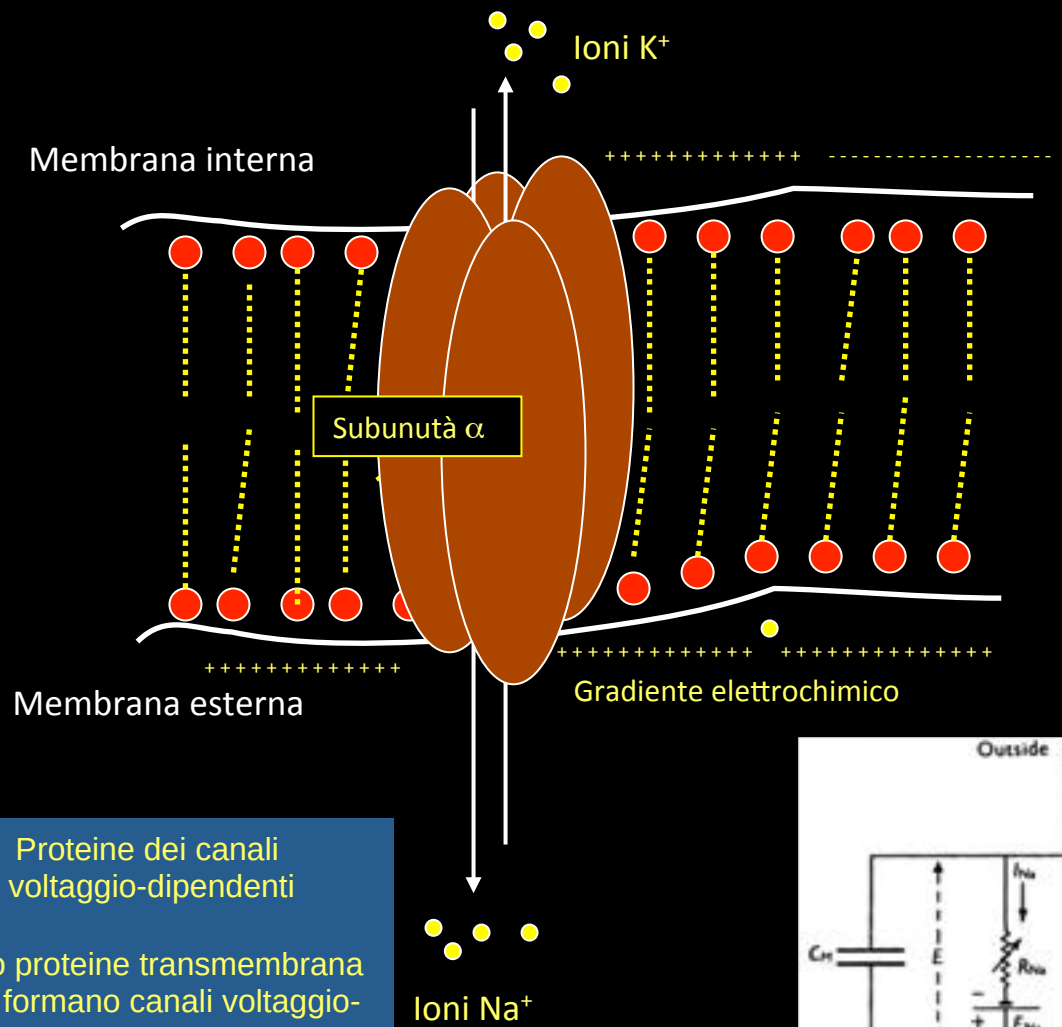
Malformazioni cranio-facciali e neurosviluppo

Martino Ruggieri



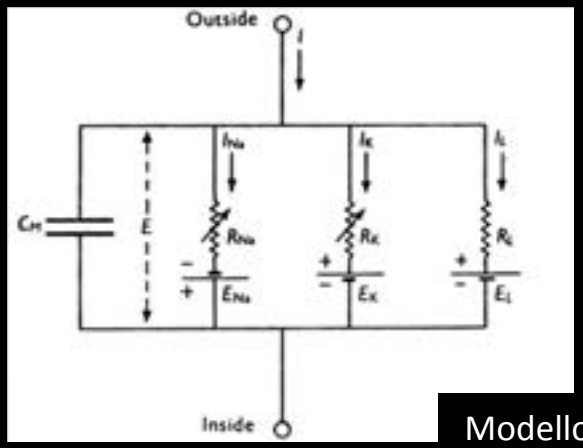
Teoria elettrica

Sodium channel, voltage-gated (brain type I) alpha subunit

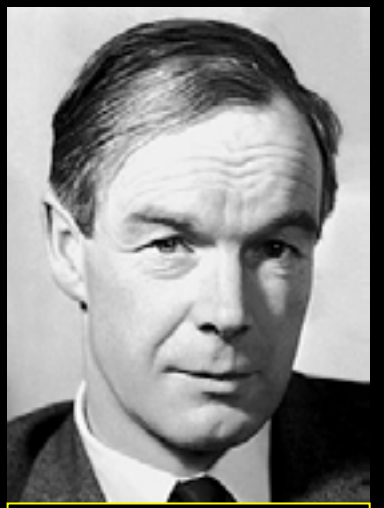


Proteine dei canali voltaggio-dipendenti

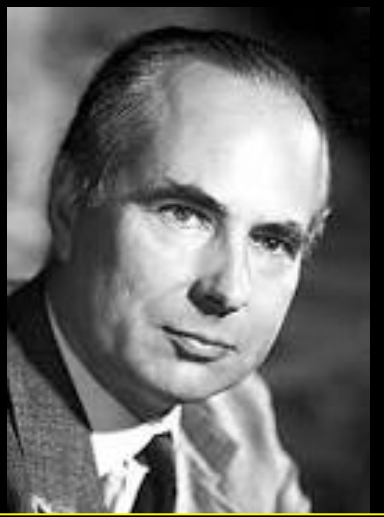
Sono proteine transmembrana che formano canali voltaggio-dipendenti essenziali per la generazione e propagazione dei potebziali d'azione nei neuroni



Modello di Hodgkin-Huxley

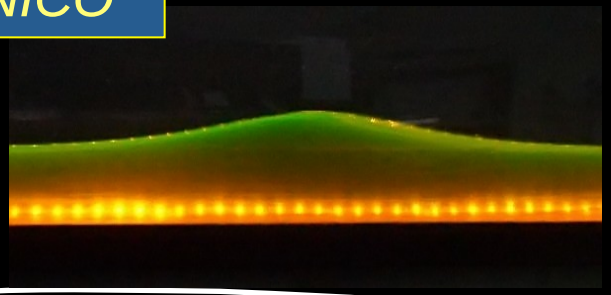


Alan Lloyd HODGKIN
[1914 - 1998]



Andrew Fielding HUXLEY
[1917 - 2012]

Teoria *meccanica → modello "SOLITONICO"*



Pulsazioni di onde sonore [SOLITONI]

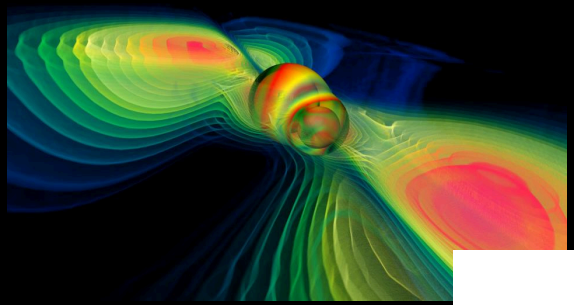
Membrana esterna

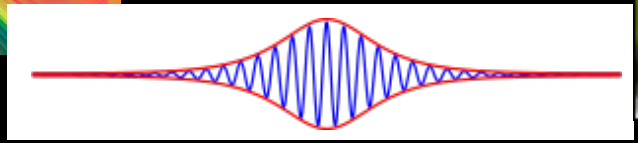
Neurone

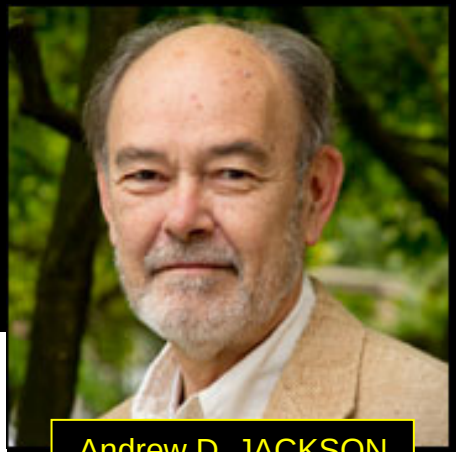
Membrana interna

Gradiente meccanico

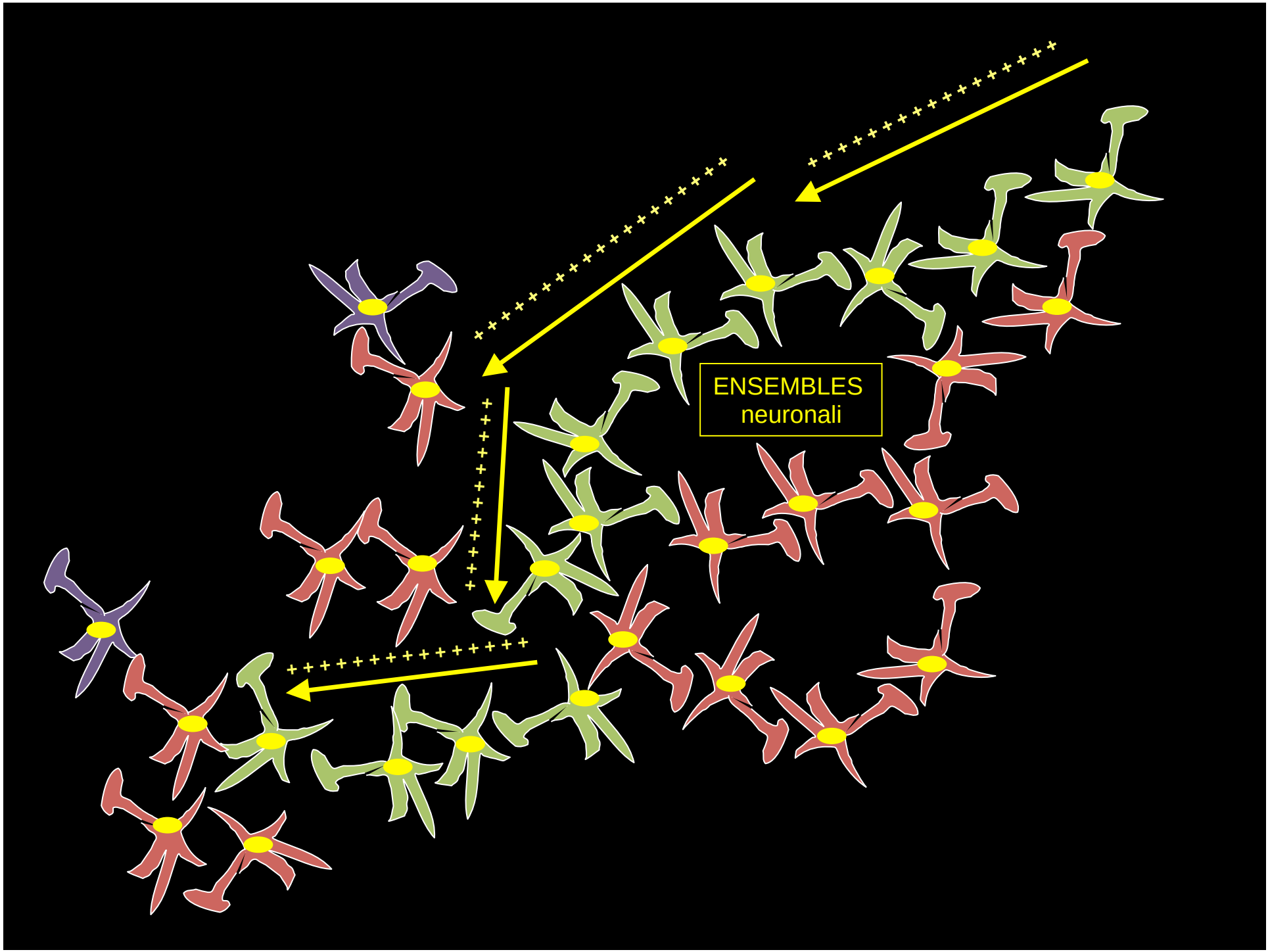
Thomas HEIMBURG

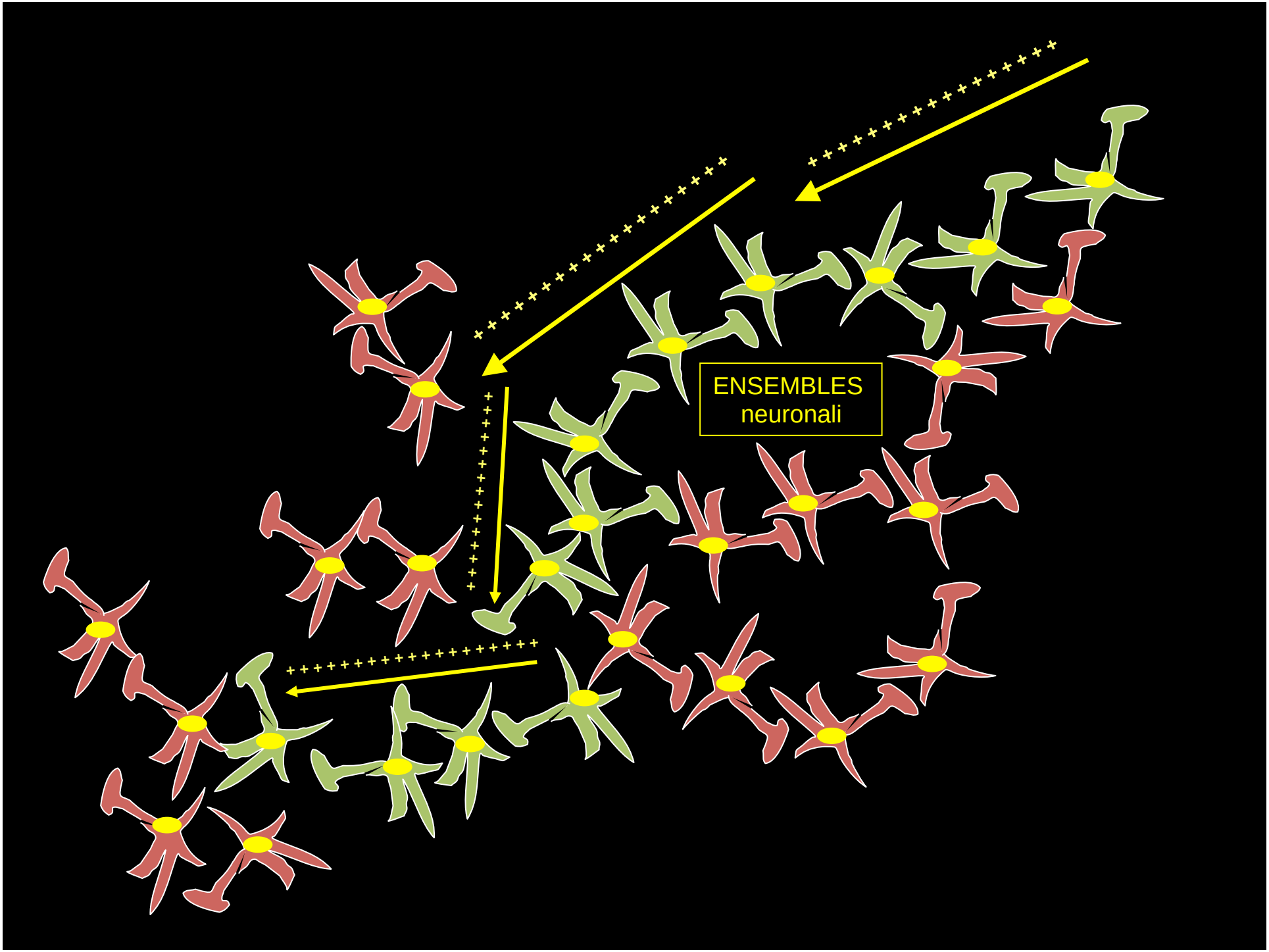


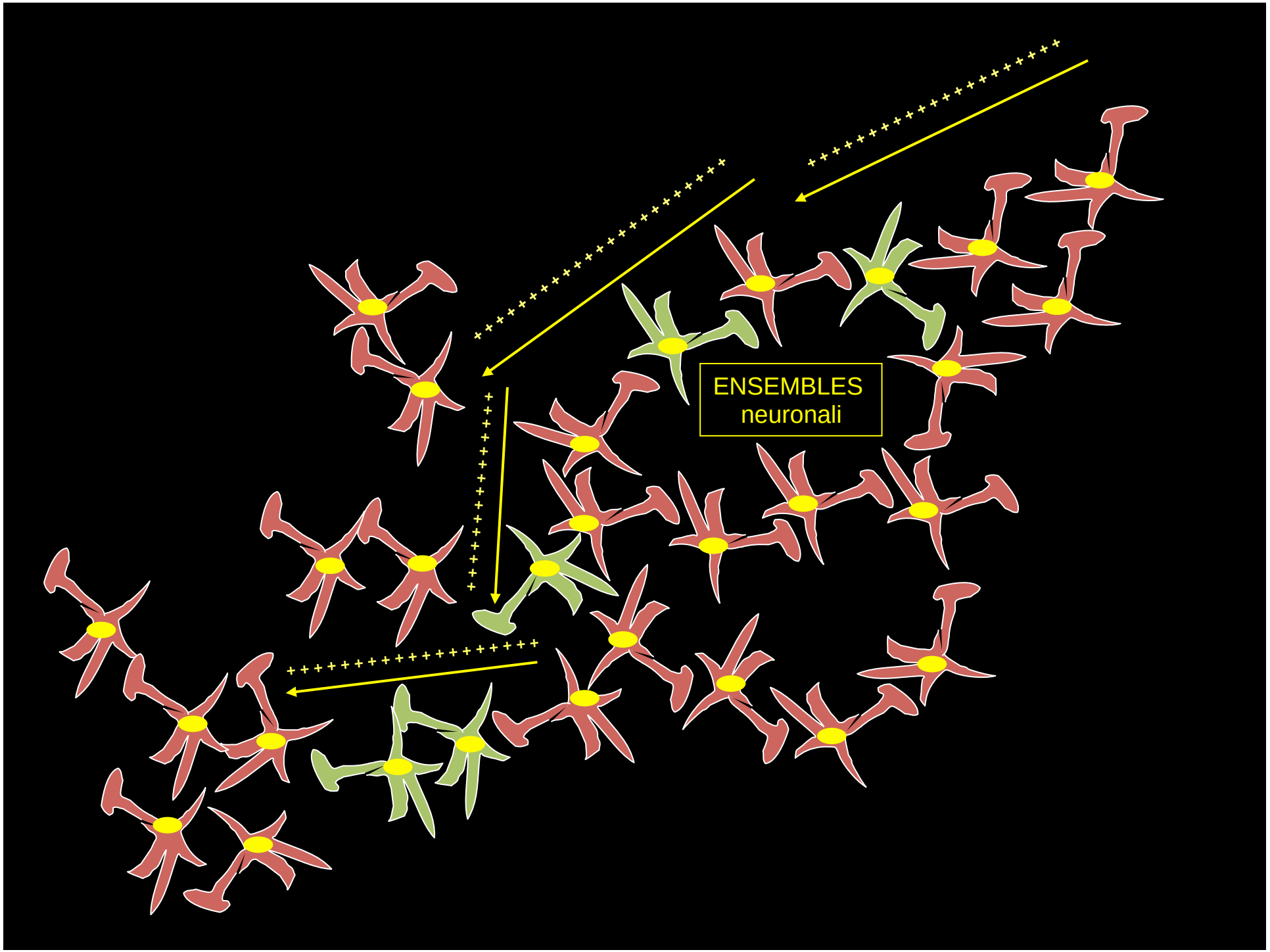


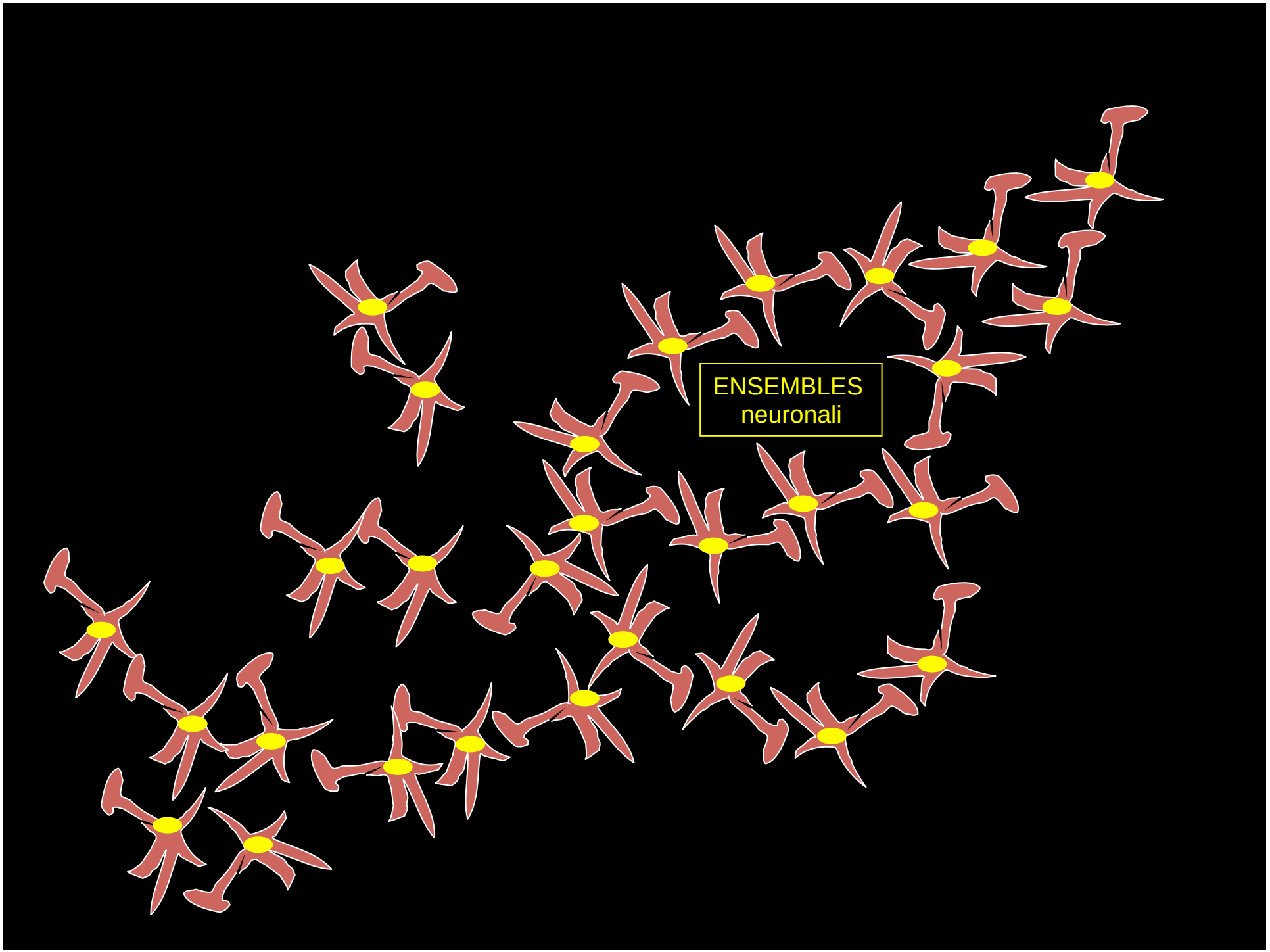


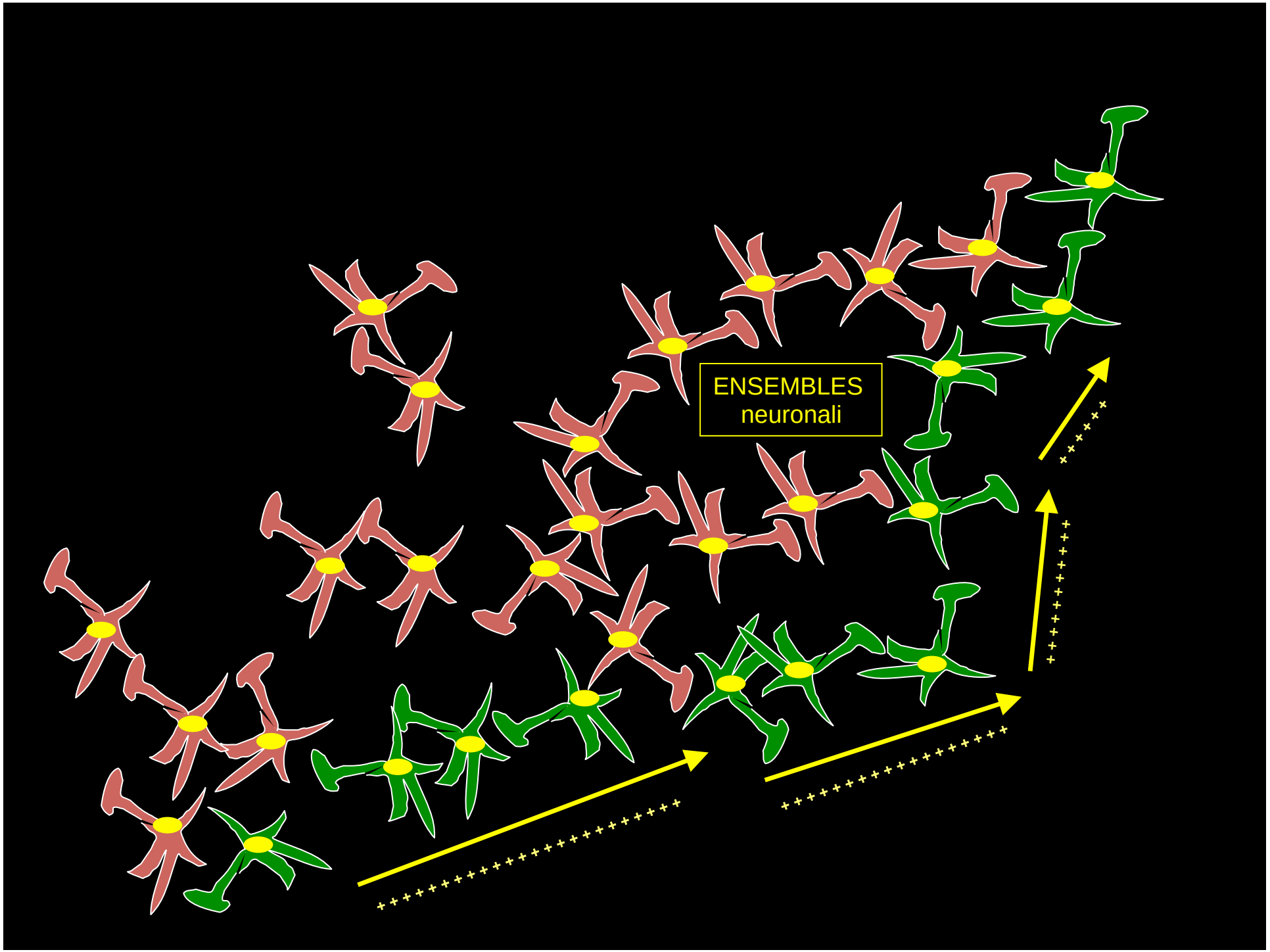
Andrew D. JACKSON

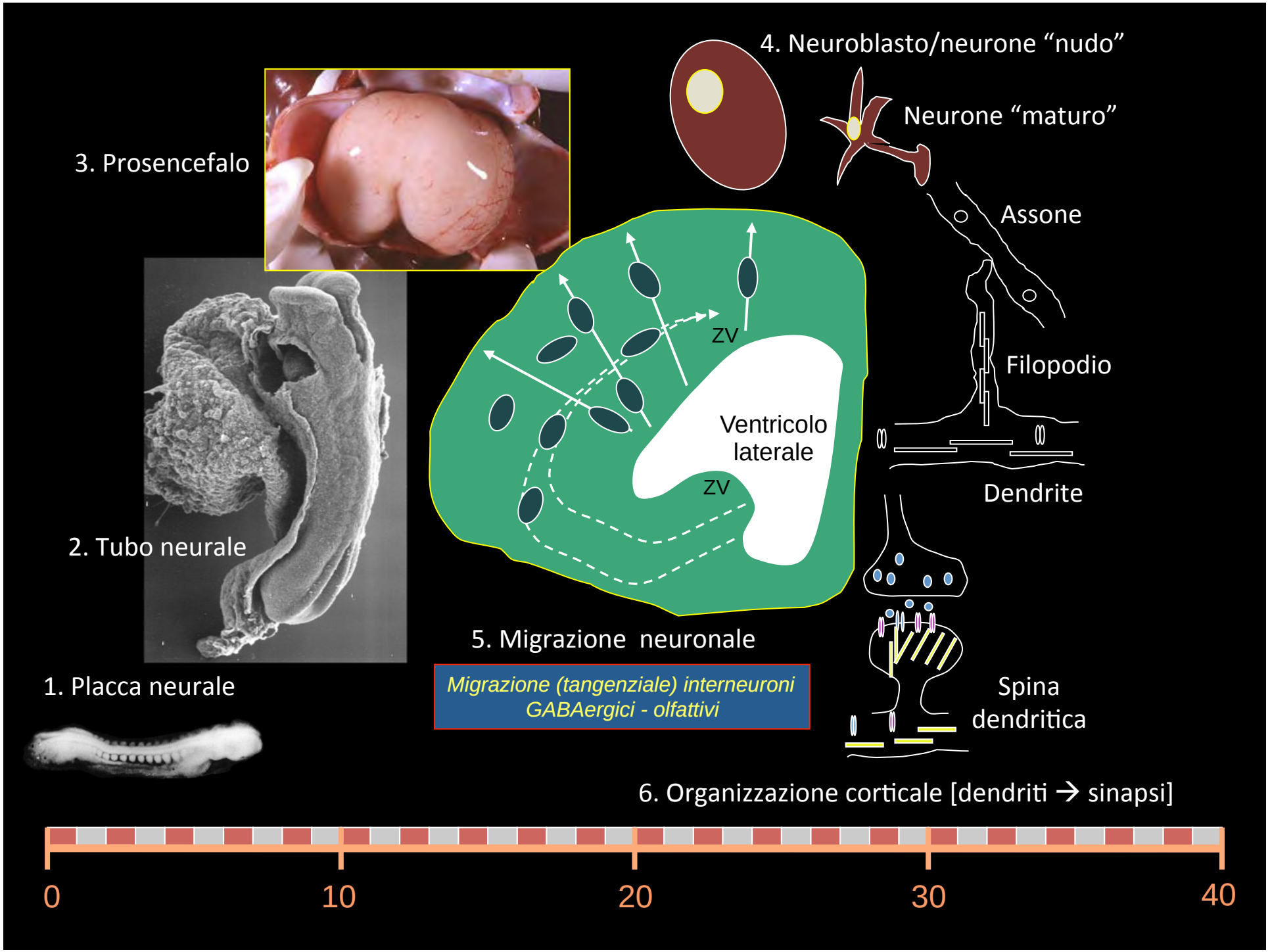

ENSEMBLES
neuronali


ENSEMBLES
neuronali

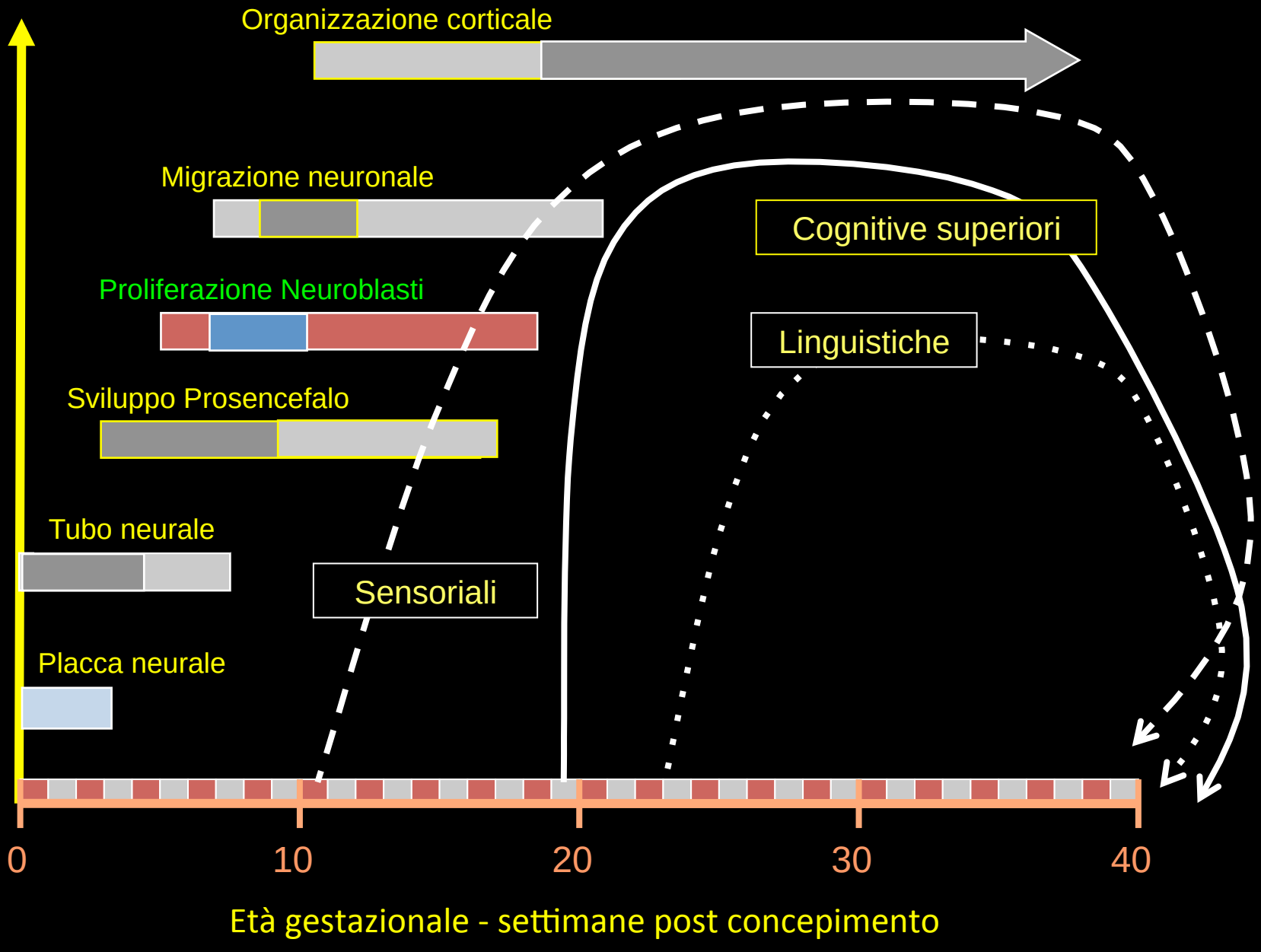

ENSEMBLES
neuronali


ENSEMBLES
neuronali

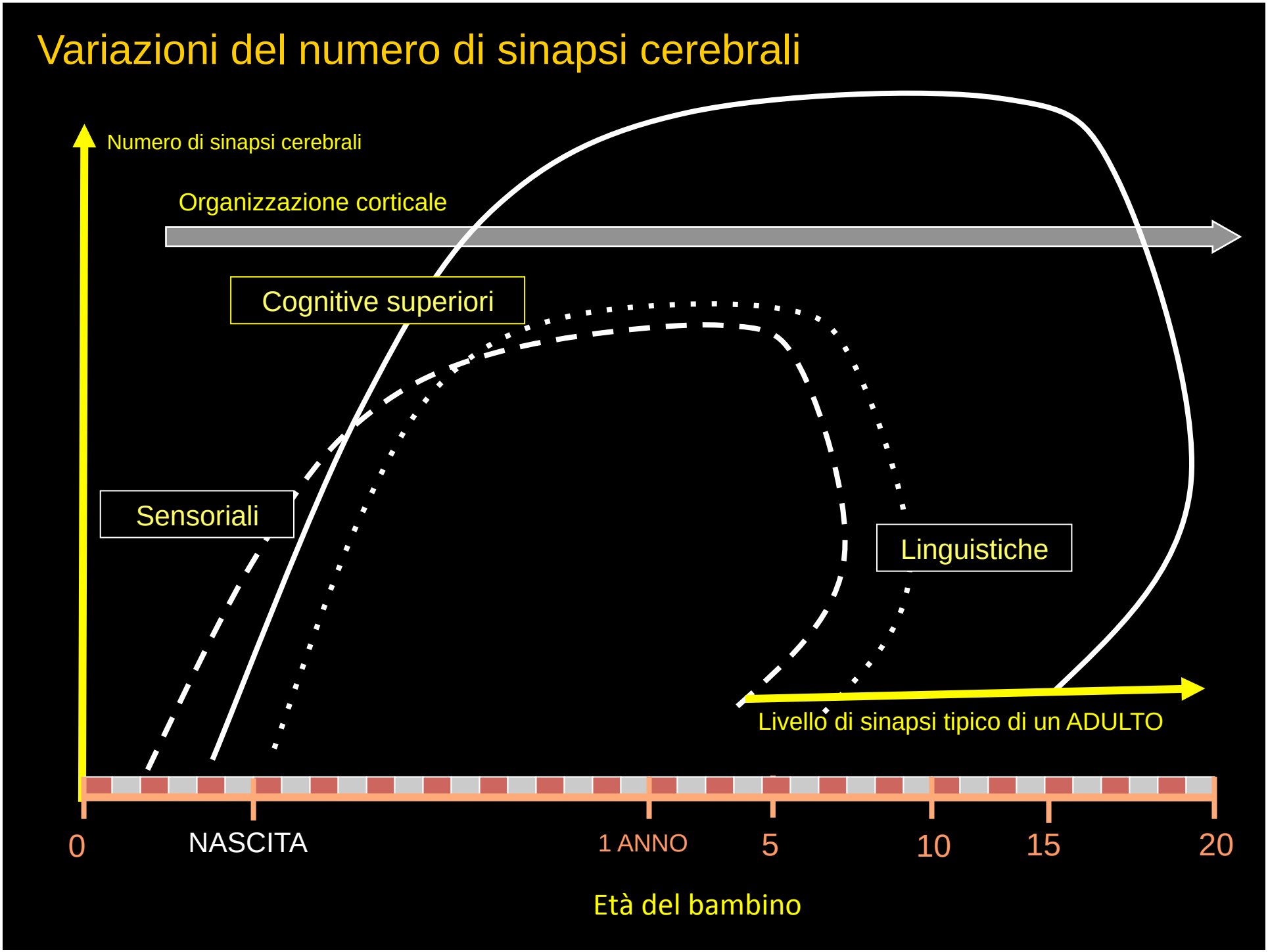

ENSEMBLES
neuronali

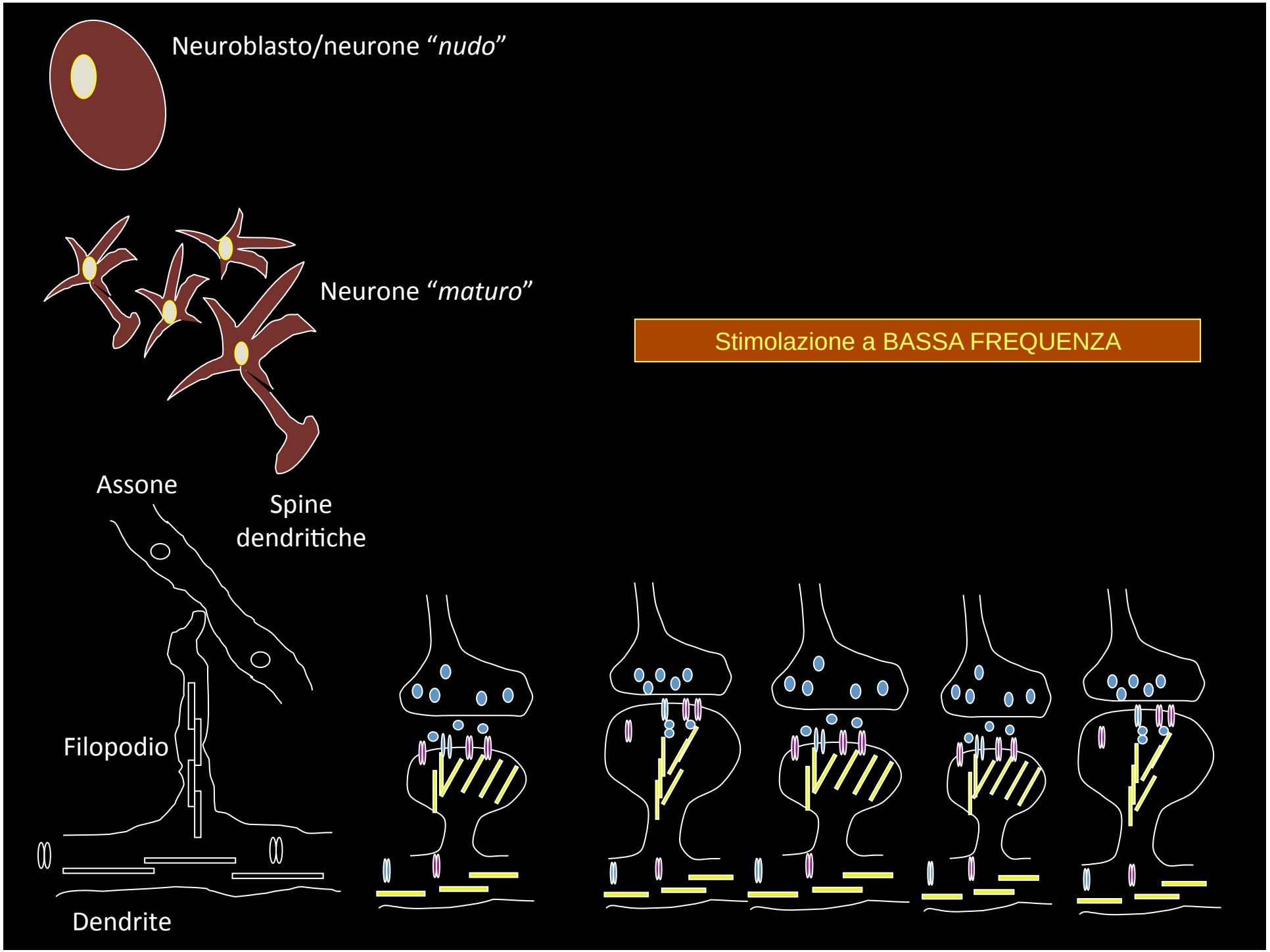

4. Neuroblasto/neurone “nudo”
Neurone “maturo”
Assone
Filopodio
Dendrite
Spina dendritica
3. Prosencefalo
2. Tubo neurale
1. Placca neurale
ZV
Ventricolo laterale
ZV
5. Migrazione neuronale
Migrazione (tangenziale) interneuroni GABAergici - olfattivi
6. Organizzazione corticale [dendriti → sinapsi]
0
10
20
30
40

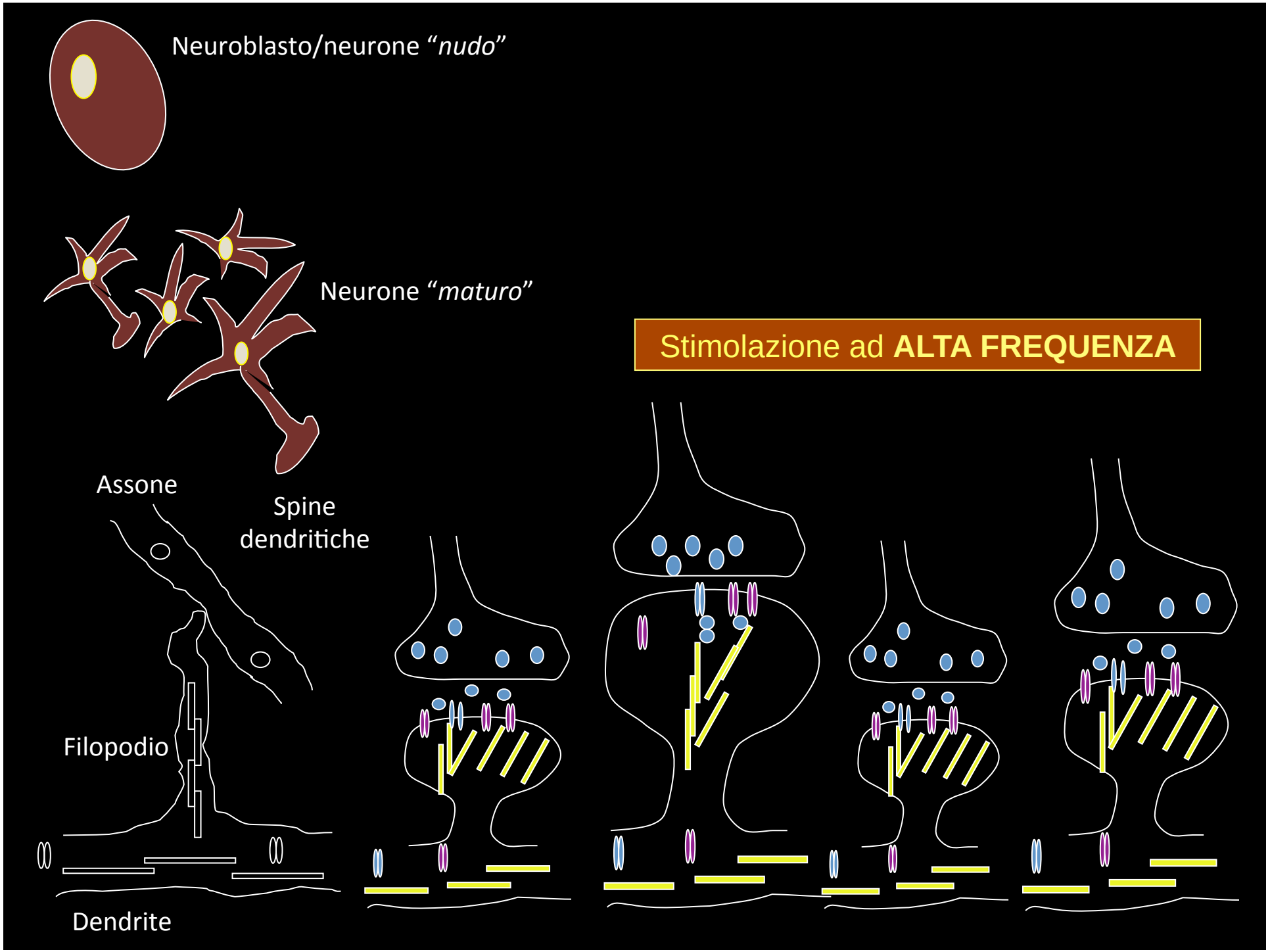
Variazioni del numero di sinapsi cerebrali



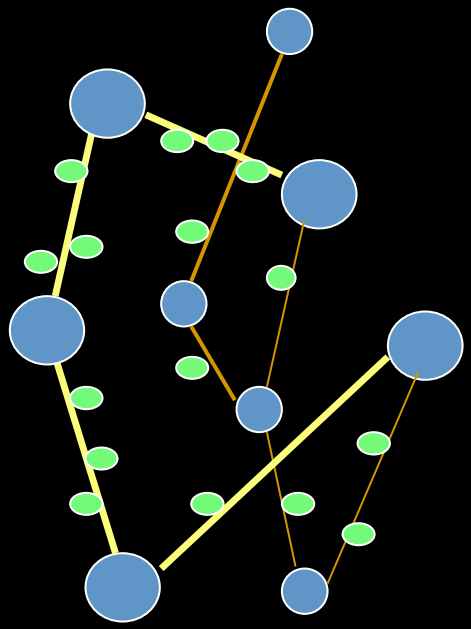
Variazioni del numero di sinapsi cerebrali



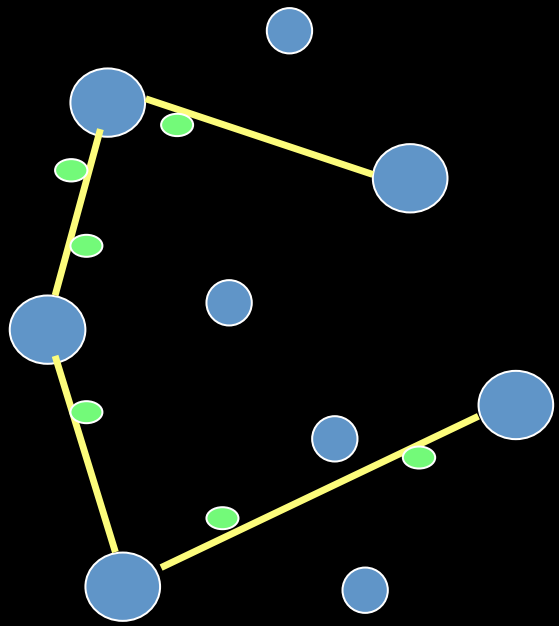

Neuroblasto/neurone “*nudo*”
Neurone “*maturo*”
Stimolazione a BASSA FREQUENZA
Assone
Spine dendritiche
Filopodio
Dendrite

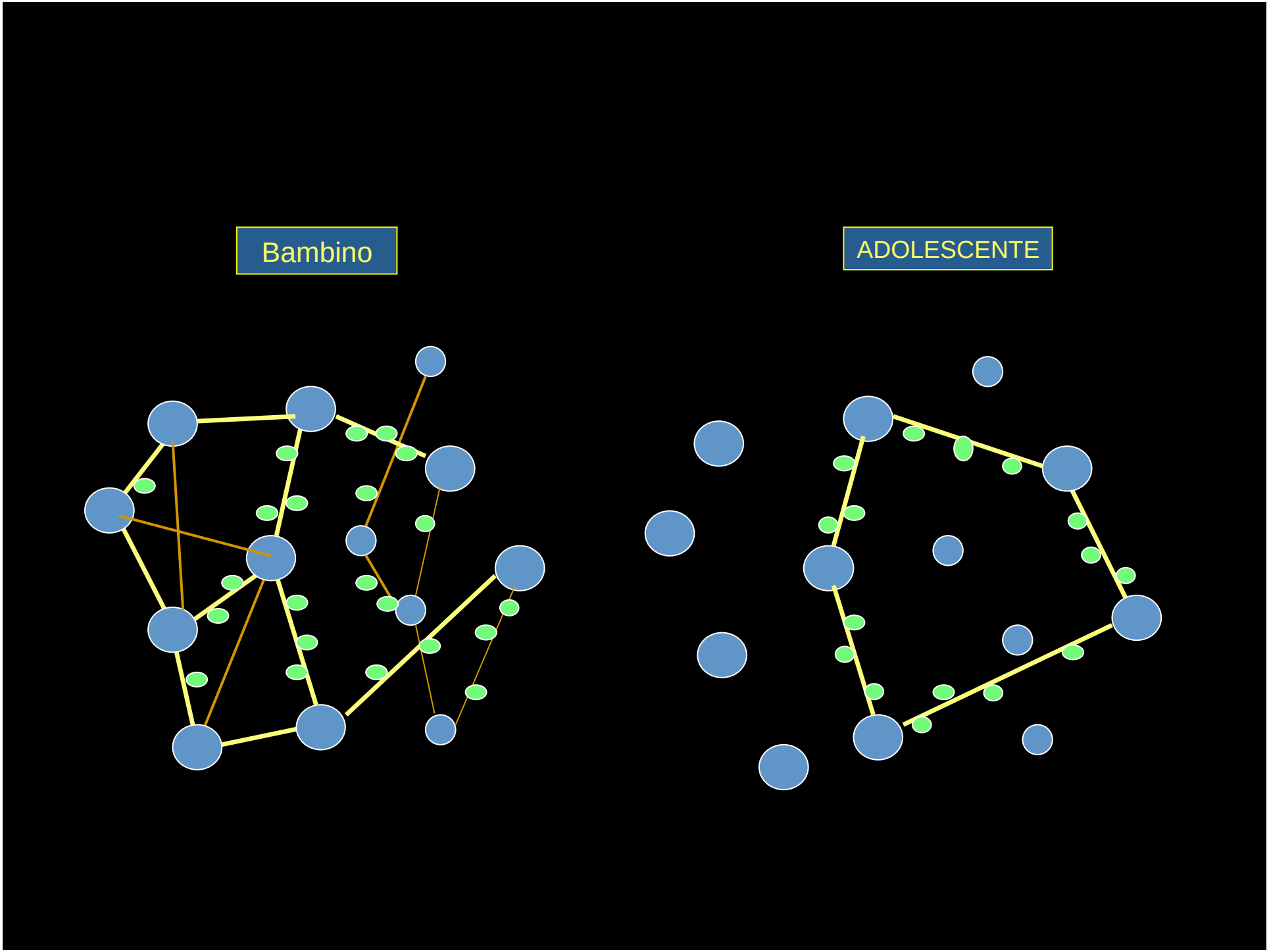

Neuroblasto/neurone “*nudo*”
Neurone “*maturo*”
Stimolazione ad **ALTA FREQUENZA**
Assone
Spine dendritiche
Filopodio
Dendrite

VEGLIA

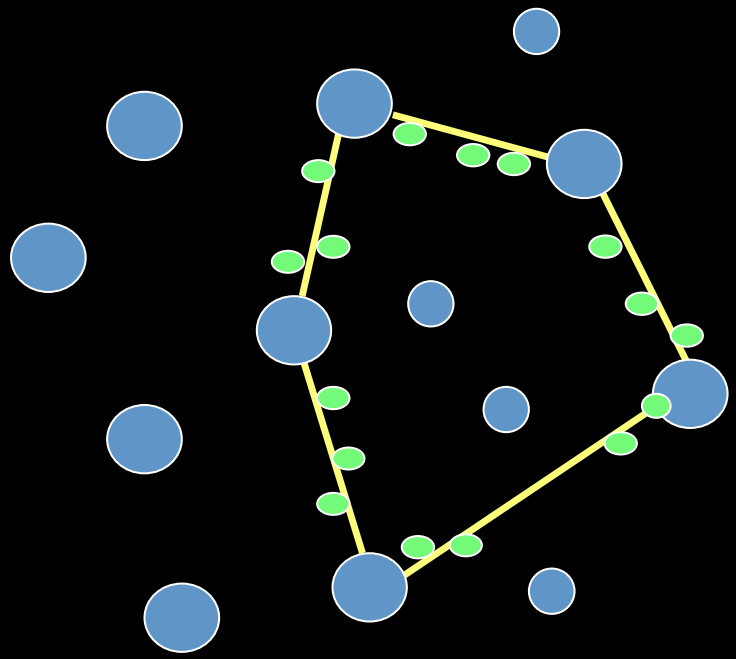


SONNO

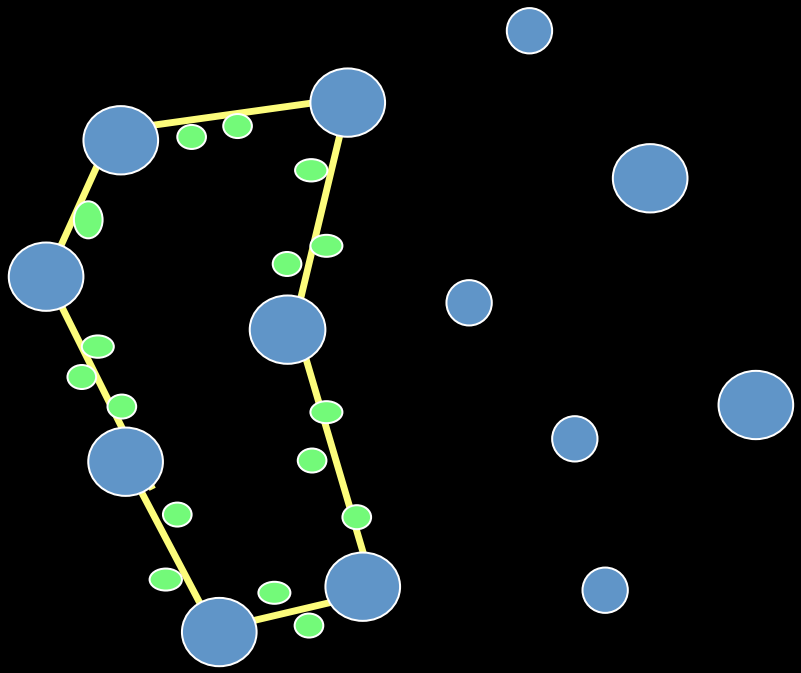


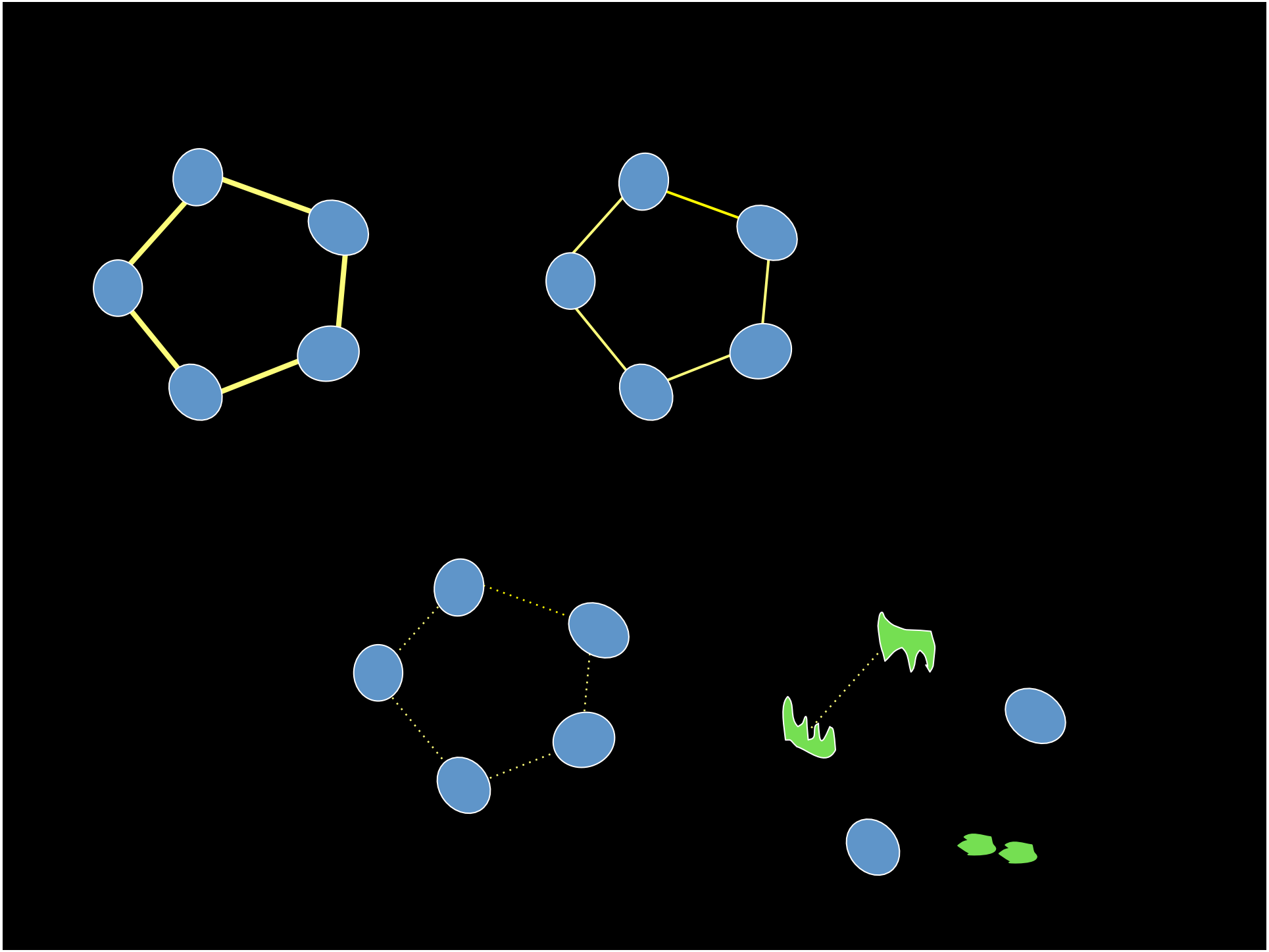

Bambino
ADOLESCENTE

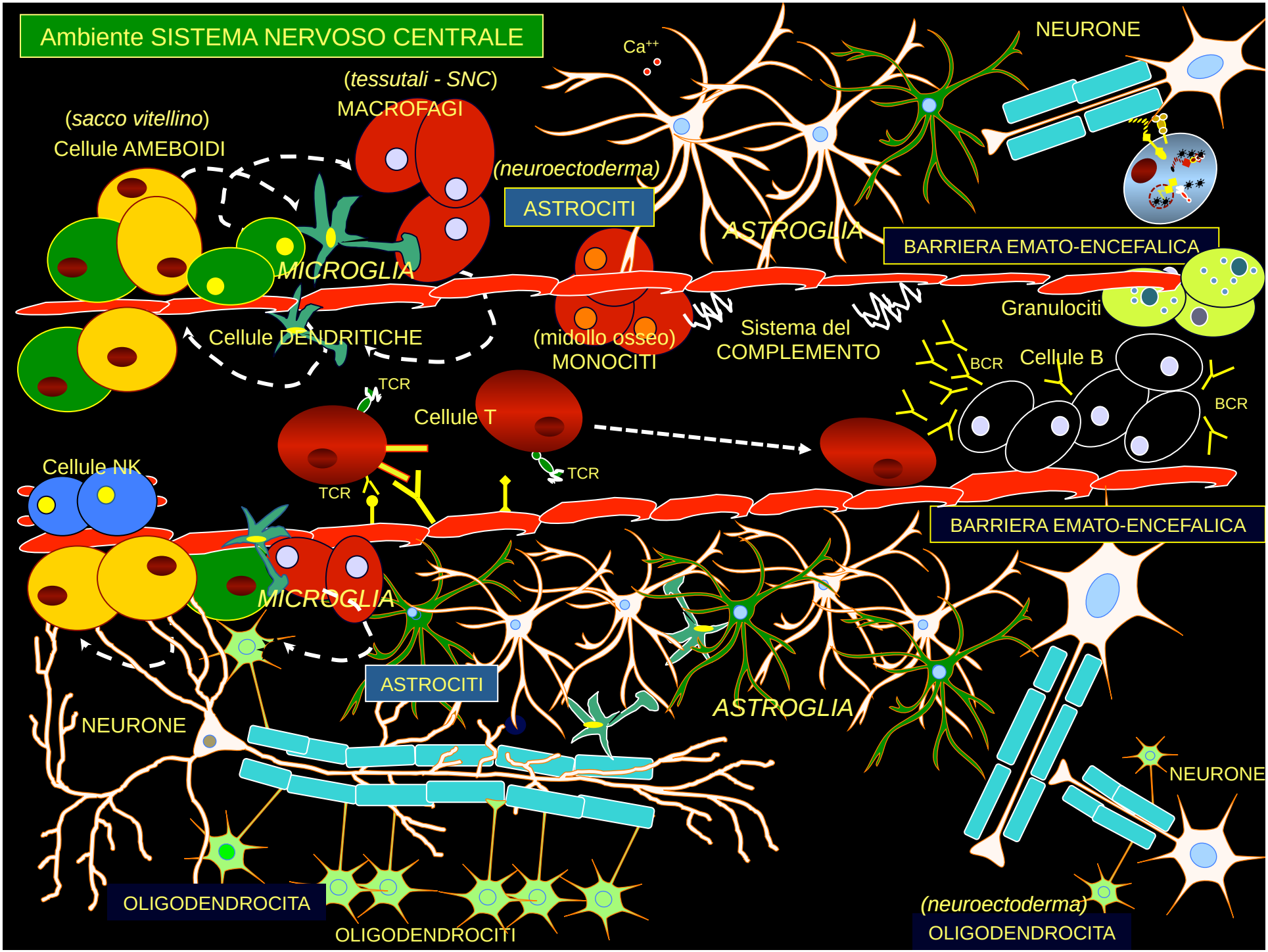
ADOLESCENTE - ***14 anni***

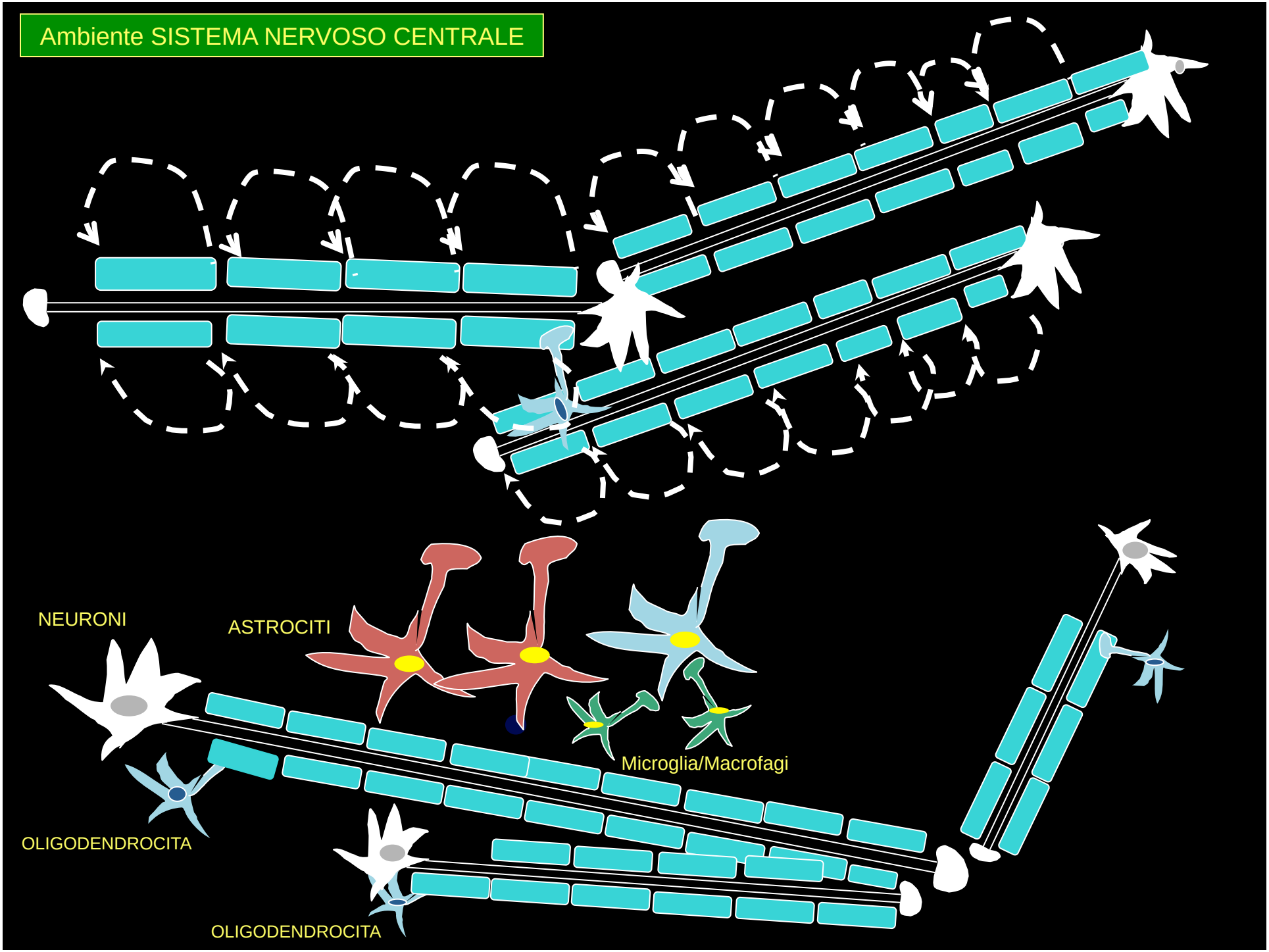


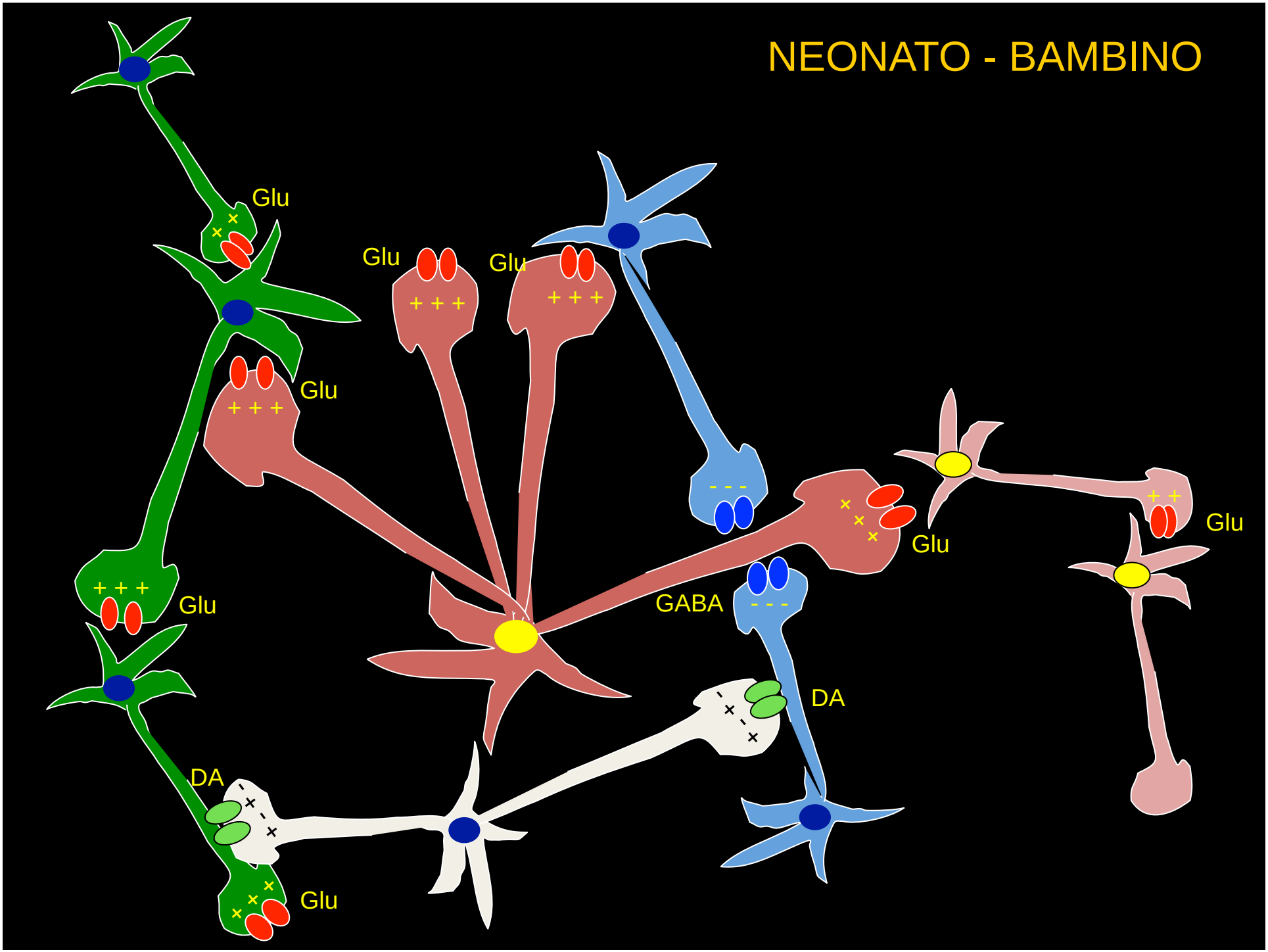
ADOLESCENTE - *22 anni*

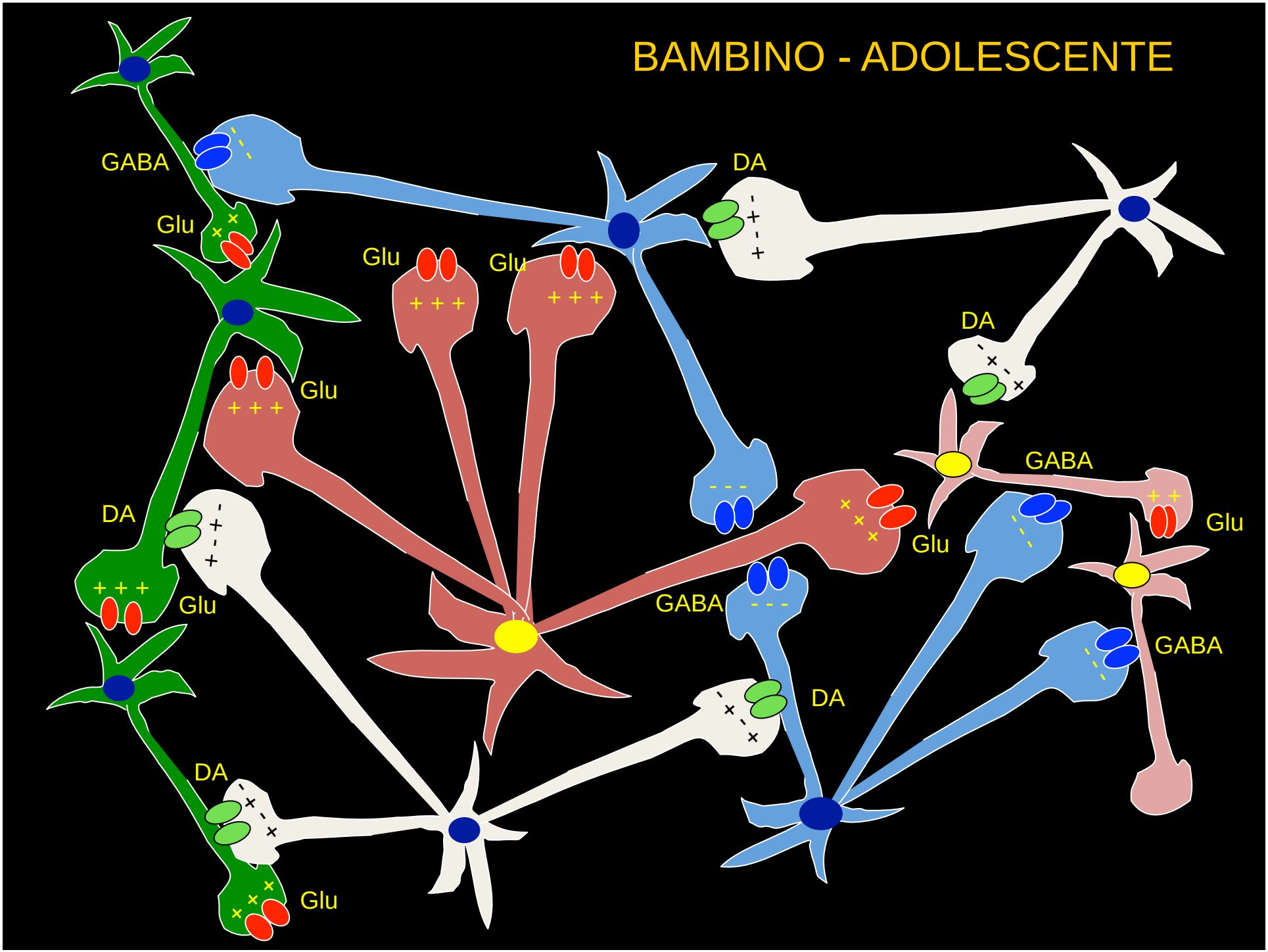


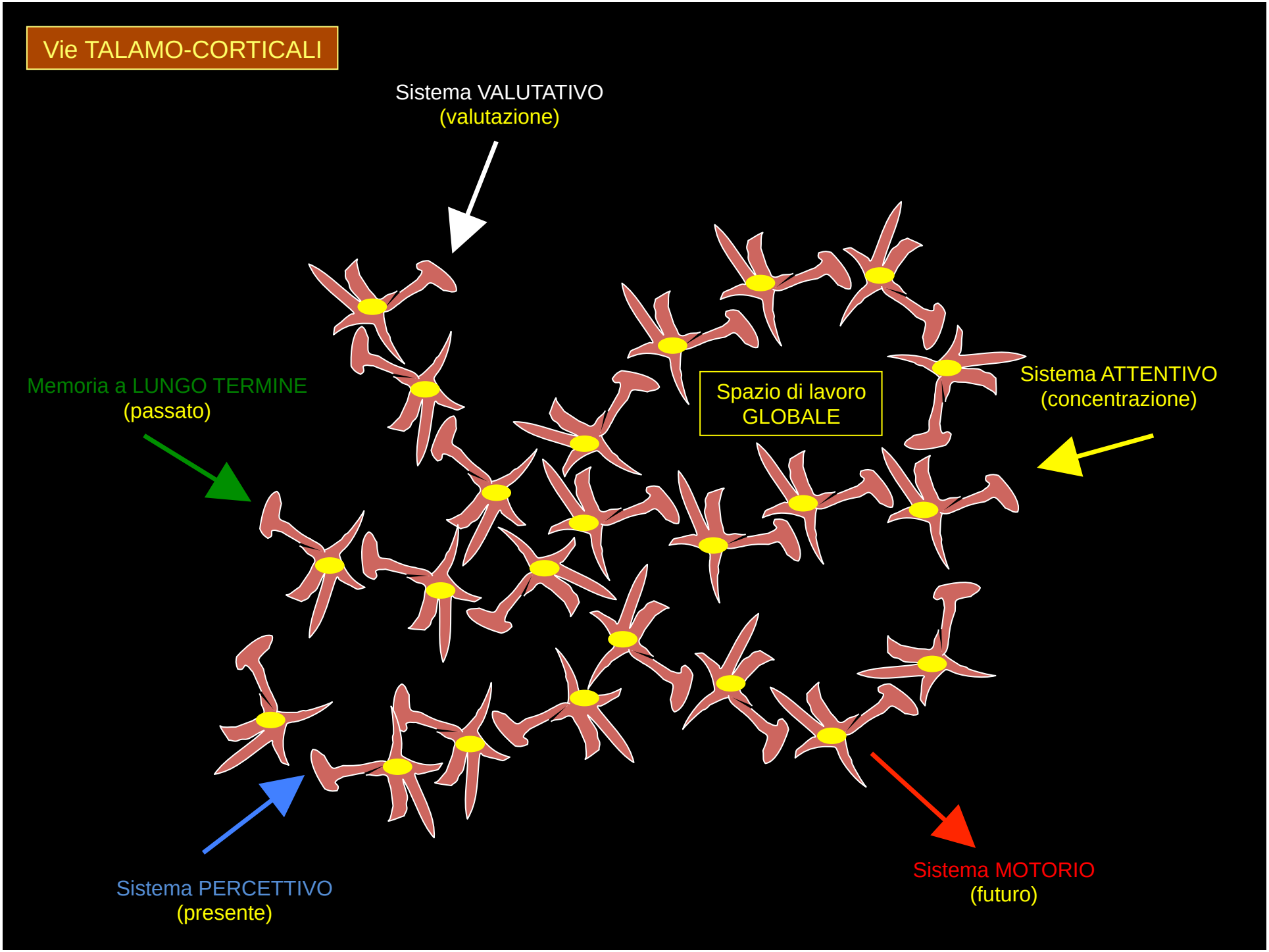



Ambiente SISTEMA NERVOSO CENTRALE
Ca++
NEURONE
(tessutali - SNC)
MACROFAGI
(sacco vitellino)
Cellule AMEBOIDI
(neuroectoderma)
ASTROCITI
ASTROGLIA
BARRIERA EMATO-ENCEFALICA
MICROGLIA
Granulociti
Cellule DENDRITICHE
(midollo osseo)
MONOCITI
Sistema del
COMPLEMENTO
BCR
Cellule B
BCR
TCR
Cellule T
TCR
TCR
Cellule NK
BARRIERA EMATO-ENCEFALICA
MICROGLIA
ASTROCITI
ASTROGLIA
NEURONE
NEURONE
OLIGODENDROCITA
OLIGODENDROCITI
(neuroectoderma)
OLIGODENDROCITA

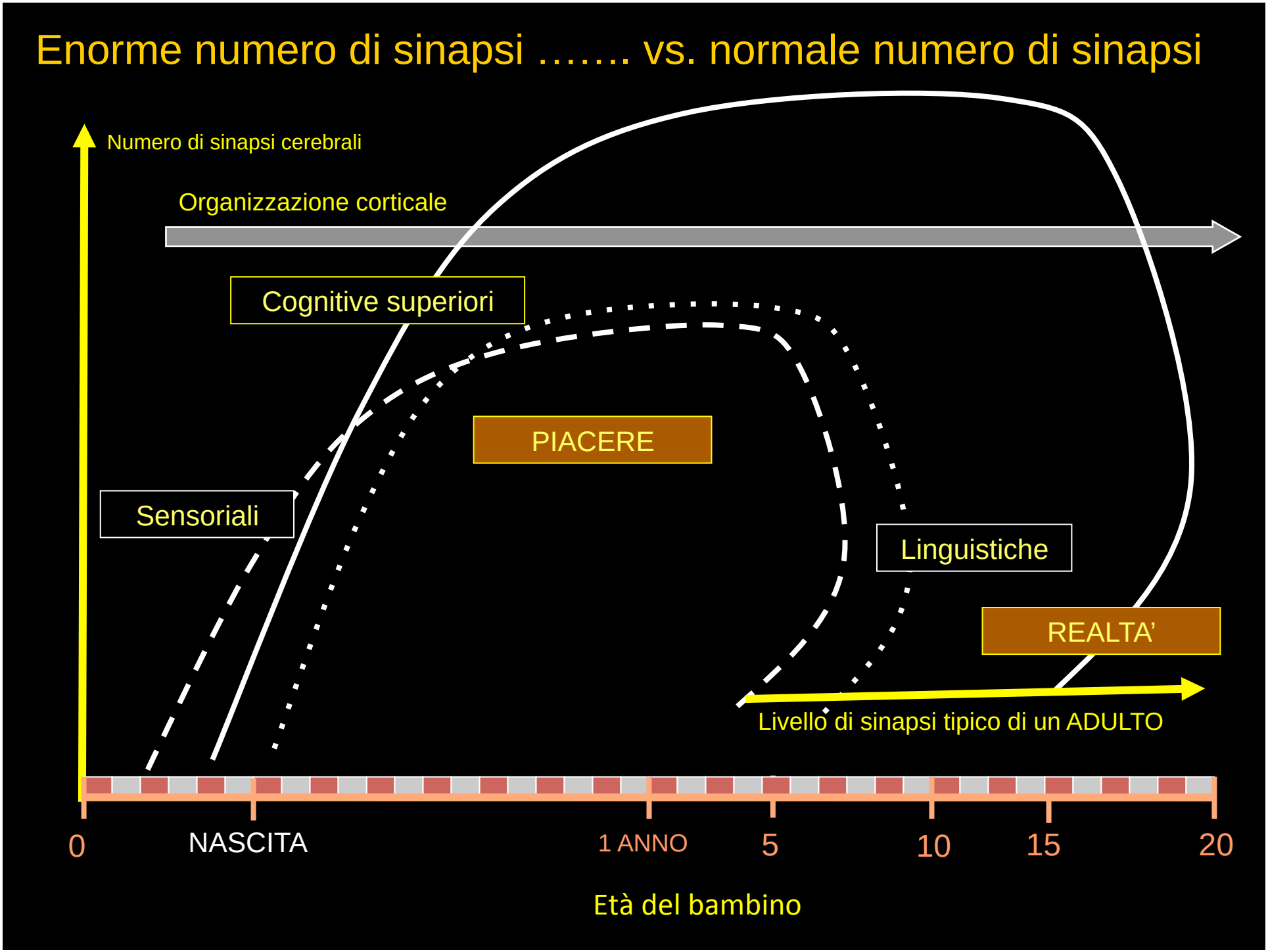

Ambiente SISTEMA NERVOSO CENTRALE
NEURONI
ASTROCITI
Microglia/Macrofagi
OLIGODENDROCITA
OLIGODENDROCITA

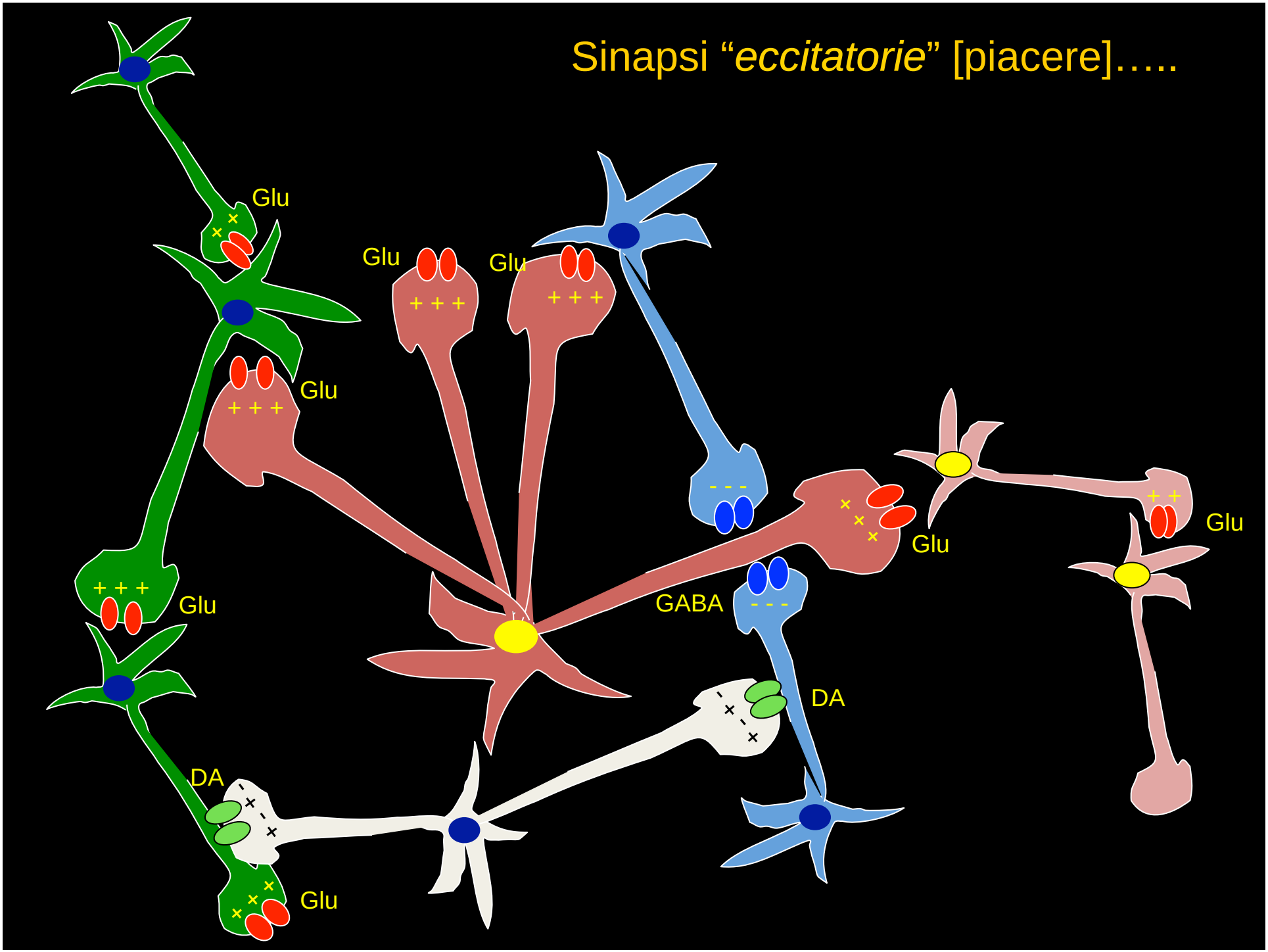

NEONATO - BAMBINO
Glu
Glu
Glu
Glu
Glu
Glu
Glu
GABA
DA
DA
Glu

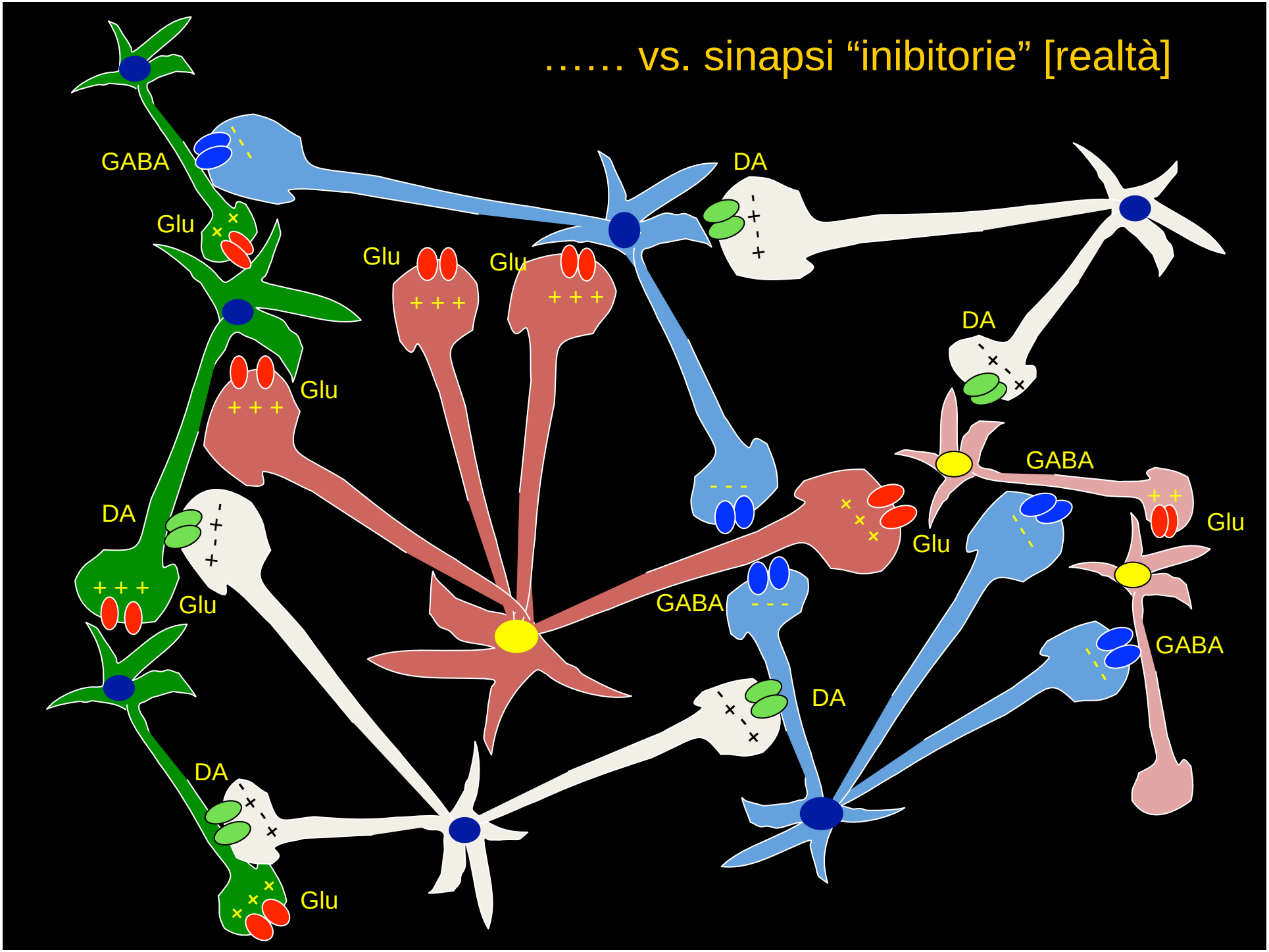

BAMBINO - ADOLESCENTE
GABA
Glu
Glu
Glu
DA
DA
Glu
DA
Glu
Glu
GABA
GABA
DA
GABA
Glu
DA
Glu


Vie TALAMO-CORTICALI
Sistema VALUTATIVO
(valutazione)
Memoria a LUNGO TERMINE
(passato)
Spazio di lavoro
GLOBALE
Sistema ATTENTIVO
(concentrazione)
Sistema PERCETTIVO
(presente)
Sistema MOTORIO
(futuro)

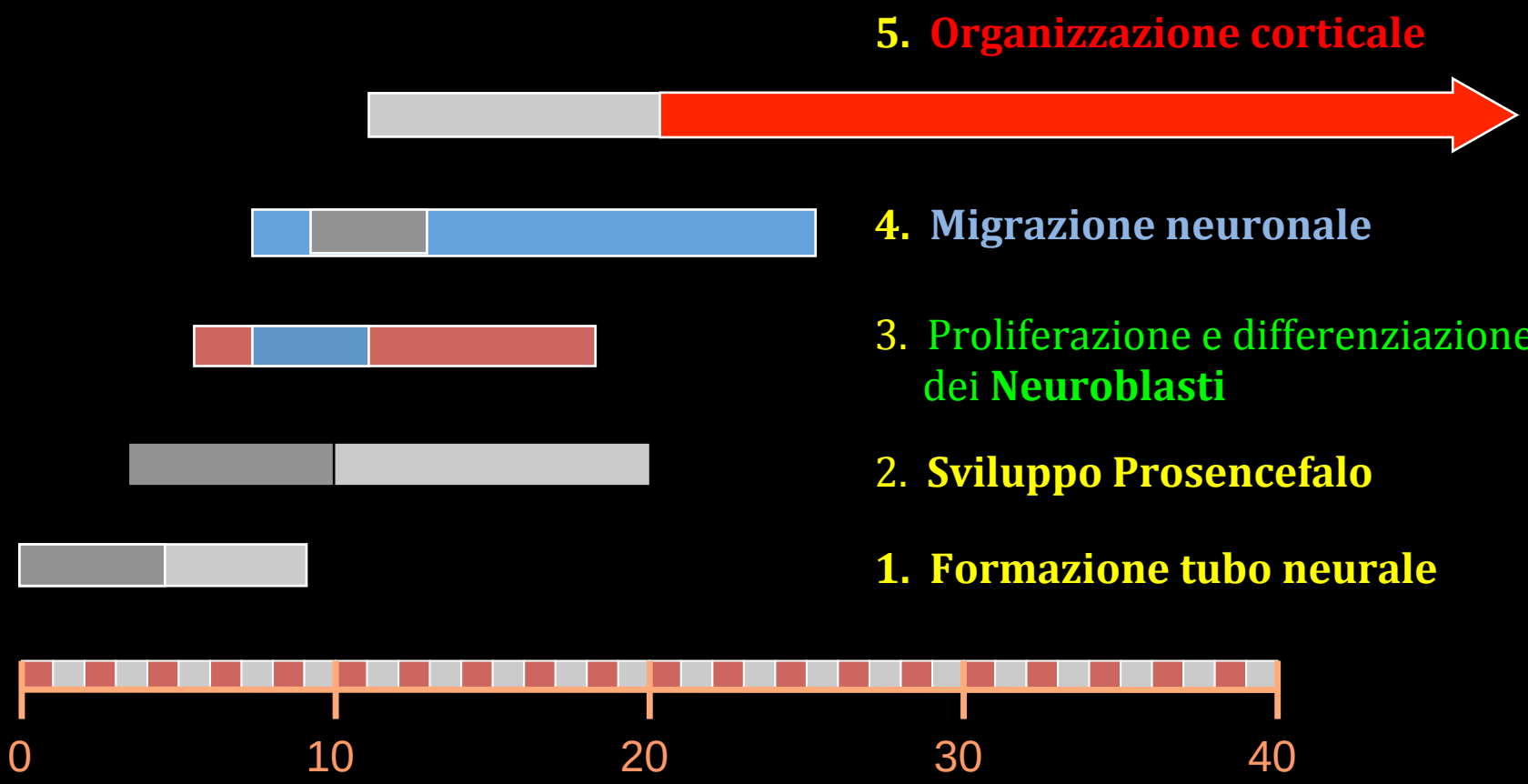
Enorme numero di sinapsi ……. vs. normale numero di sinapsi




Sinapsi “*eccitatorie*” [piacere]…..
Glu
Glu
Glu
Glu
+ + +
Glu
+ + +
+ + +
Glu
GABA
- - -
DA
DA
Glu
- - -
+ +
Glu

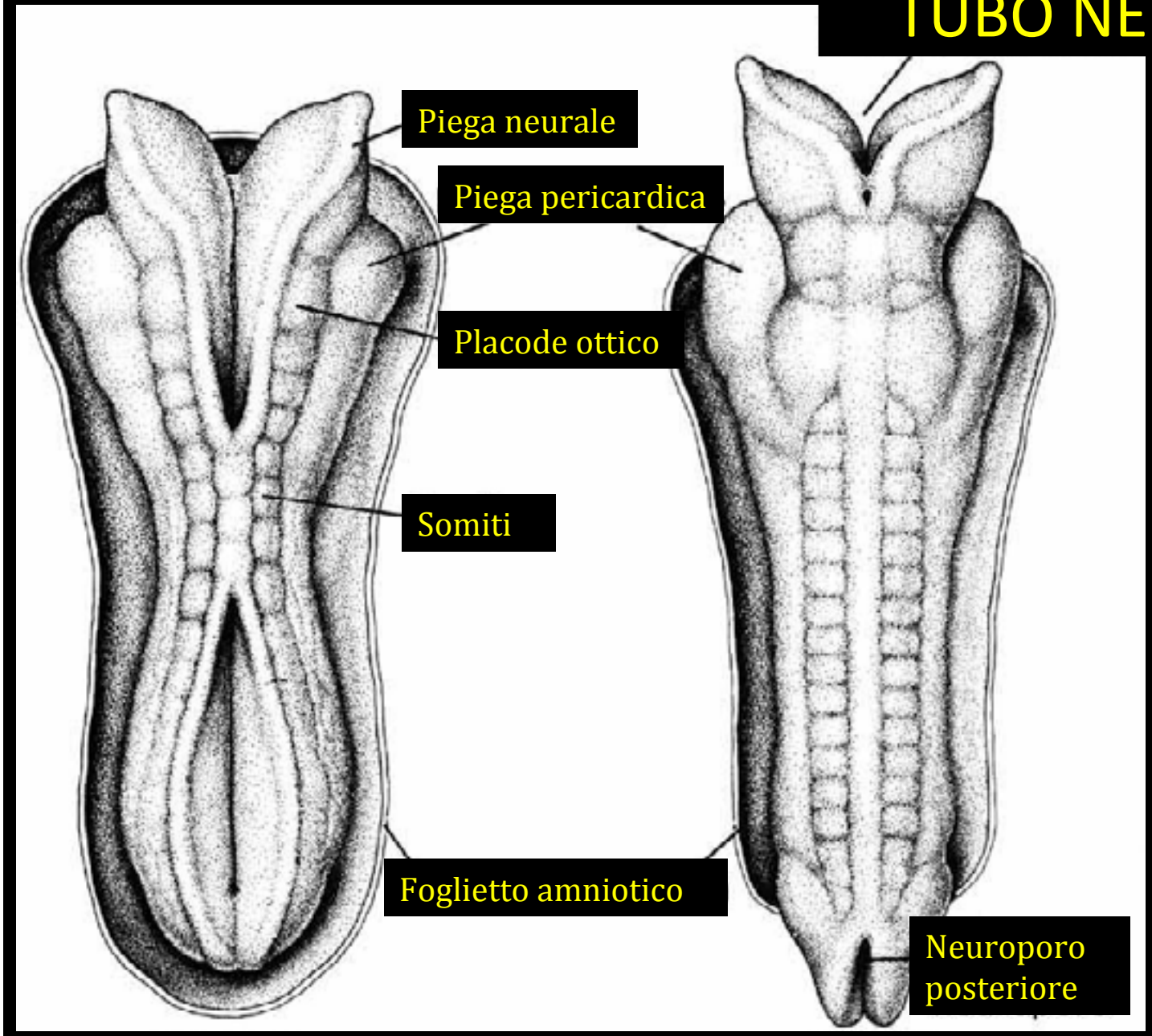

…… vs. sinapsi “inibitorie” [realtà]
GABA
Glu
Glu
Glu
DA
Glu
DA
GABA
Glu
DA
Glu
GABA
DA
GABA
DA
Glu

Sviluppo cerebrale

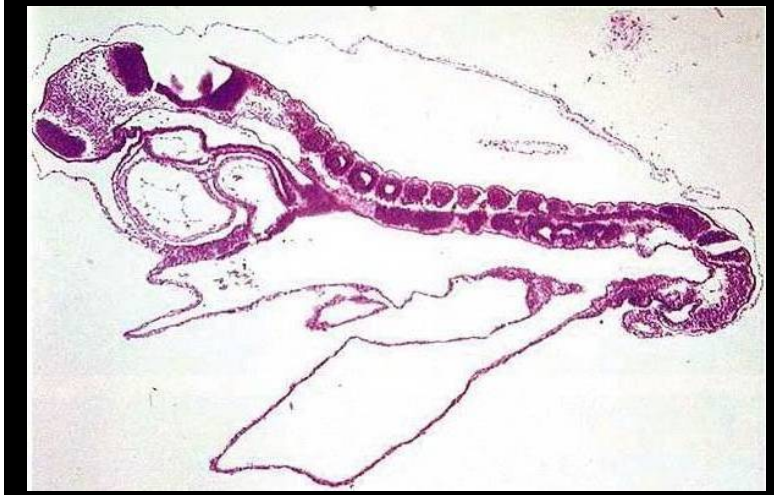


Età gestazionale - settimane post concepimento

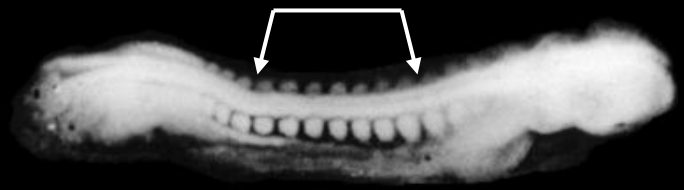
TUBO NEURALE



TUBO NEURALE

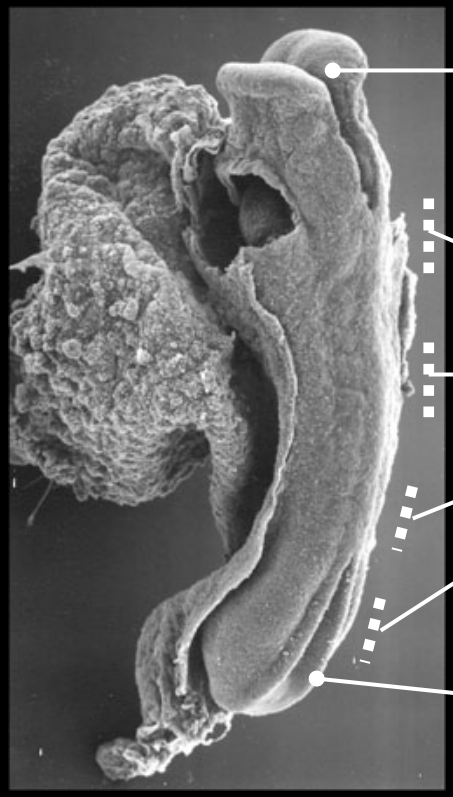


PLACCA NEURALE



15° - 16° girono di vita gestazionale

Ripiegamento e *formazione* del TUBO NEURALE



Neuroporo anteriore

Chiusura a CERNIERA multipla

Neuroporo posteriore

18° - 20° girono di vita gestazionale

DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

ANENCEFALIA

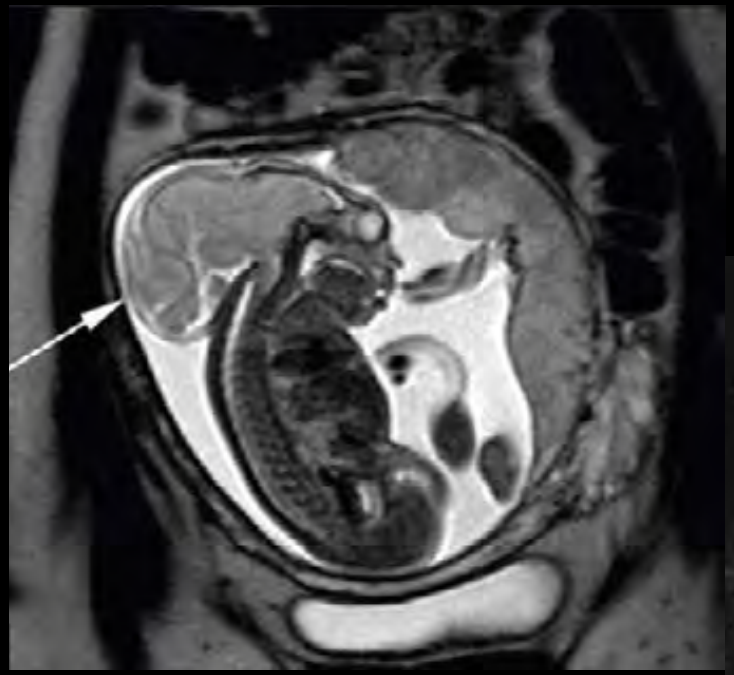
Assenza totale di tessuto cerebrale

Assenza “quasi” totale di tessuto cerebrale

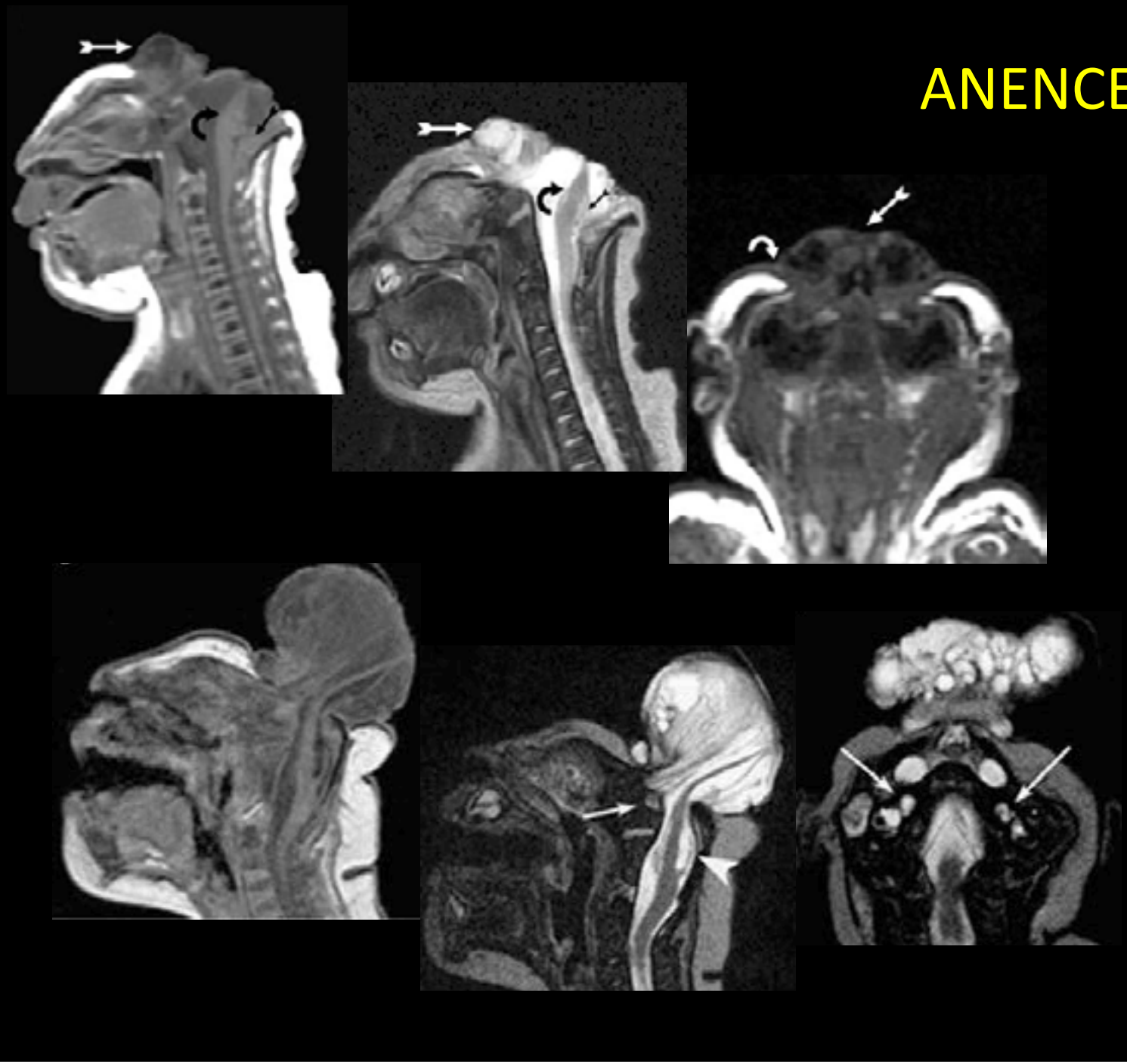
“*Area cerebrovasculosa*”
Vasi neoformati in risposta alla grave distruzione cerebrale + connettivo + glia + neuroni occasionali + ependima + plessi

ANENCEFALIA

ANENCEFALIA



ANENCEFALIA



DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

INIENCEFALIA ed ENCEFALOSCHISI (exencefalia)

ENCEFALOSCHISI

Erniazione tessuto cerebrale non formato

Mancata chiusura neuroporo anteriore

Erniazione "*area cerebrovasculosa*"

INIENCEFALIA

Retroflessione midollo spinale cervicale

Mancata formazione osso occipitale

ENCEFALOSCHISI

INIENCEFALIA

ENCEFALOSCHISI

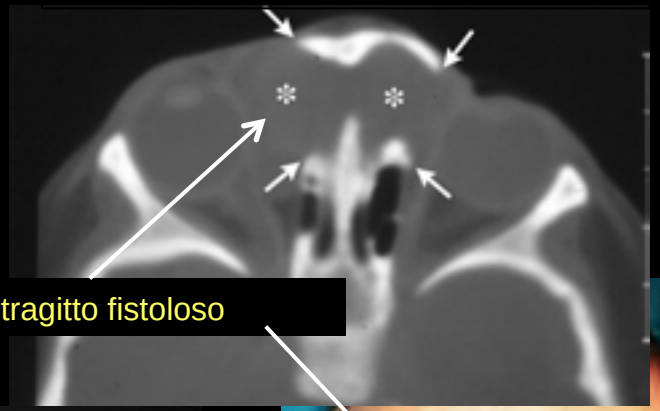
INIENCEFALIA

DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

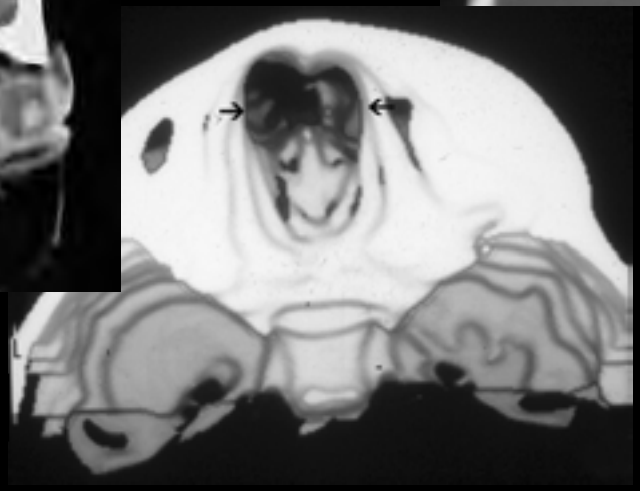
CEFALOCELE

Protrusione delle strutture intracraniche [encefaliche (ENCEFALOCELE) e/o meningee (MENINGOCELE) attraverso difetti delle ossa del cranio e della dura madre

CEFALOCELE NASOFARINGEO

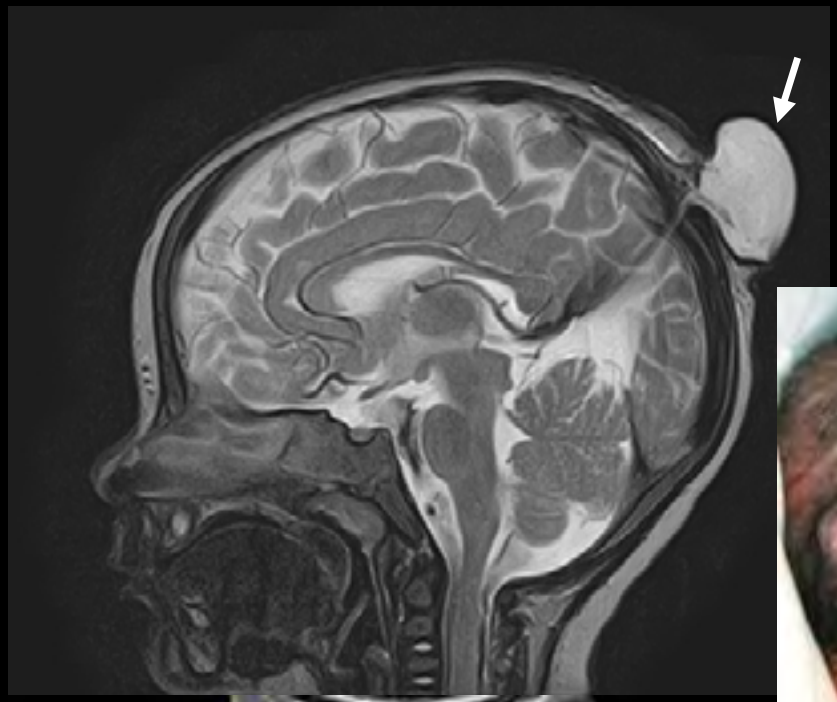


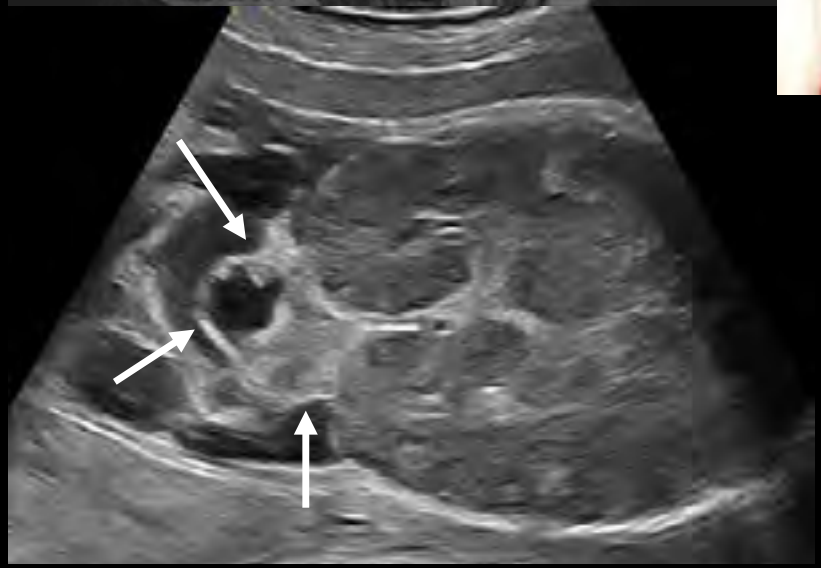
Mancata regressione proiezione dura madre nasale con creazione tragitto fistoloso



CEFALOCELE FRONTOETMOIDALE

CEFALOCELE





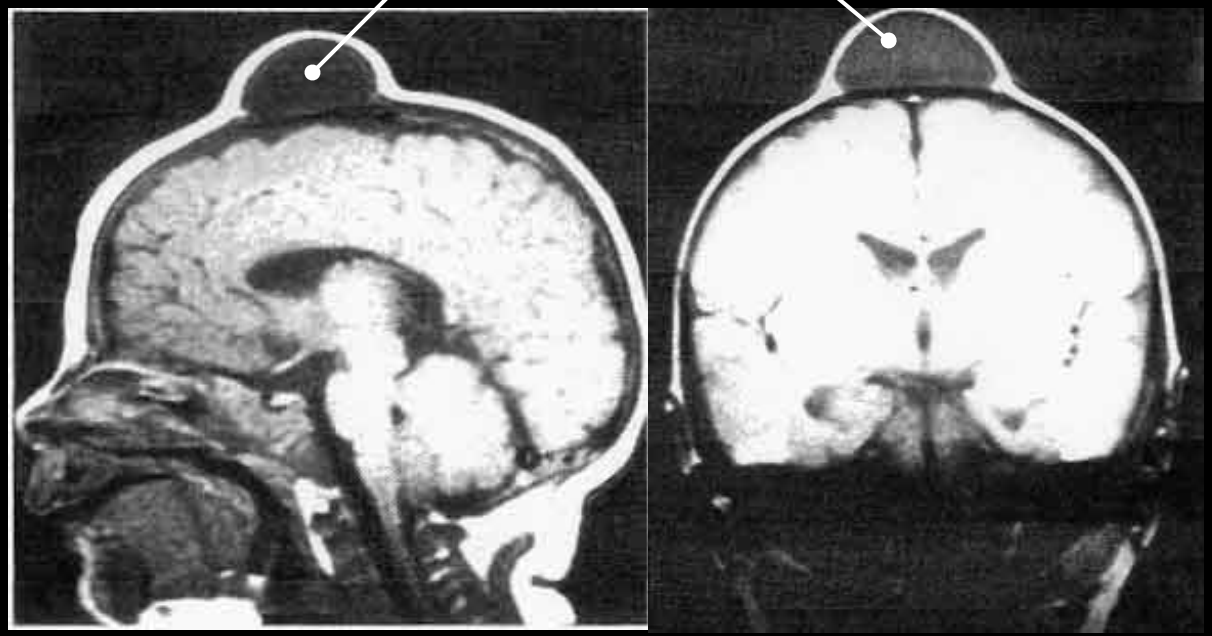
DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

CEFALOCELE

Protrusione delle strutture intracraniche [encefaliche (ENCEFALOCELE) e/o meningee (MENINGOCELE)] attraverso difetti delle ossa del cranio e della dura

CEFALOCELI della CONVESSITA'

Cefalocele sagittale



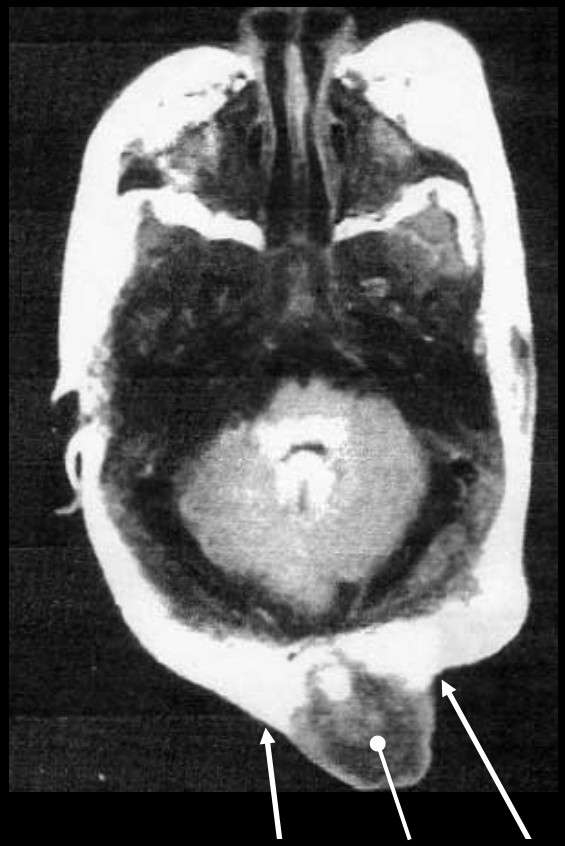
Cefalocele occipitocervicale

- occipito-cervicali
- occipitali
- sagittali o parietali
- laterali
- bregmatici
- interfrontali
- temporali

DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

CEFALOCELE ATRESICO

Erniazione di DURA MADRE e TESSUTO FIBROSO (in associazione a tessuto cerebrale degenerato) attraverso le ossa della regione parieto-occipitale



Dura madre + tessuto fibroso + tessuto cerebrale degenerato

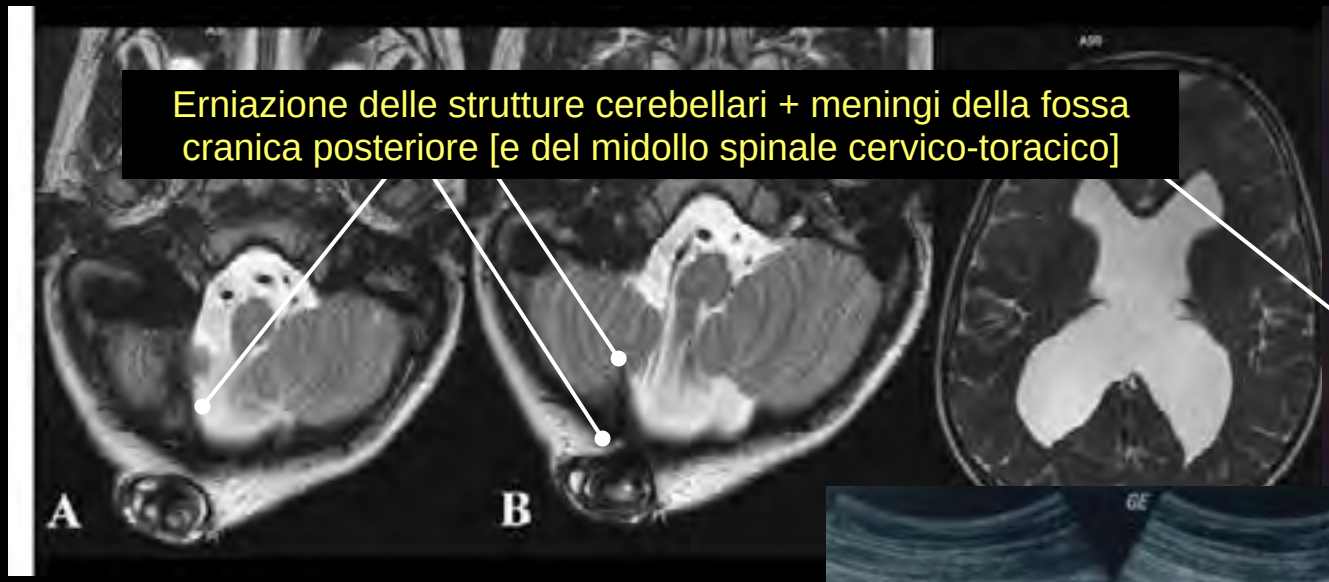
Meningoencefalocele anteriore *fronto-nasale*

Bambino con liquorrea nasale

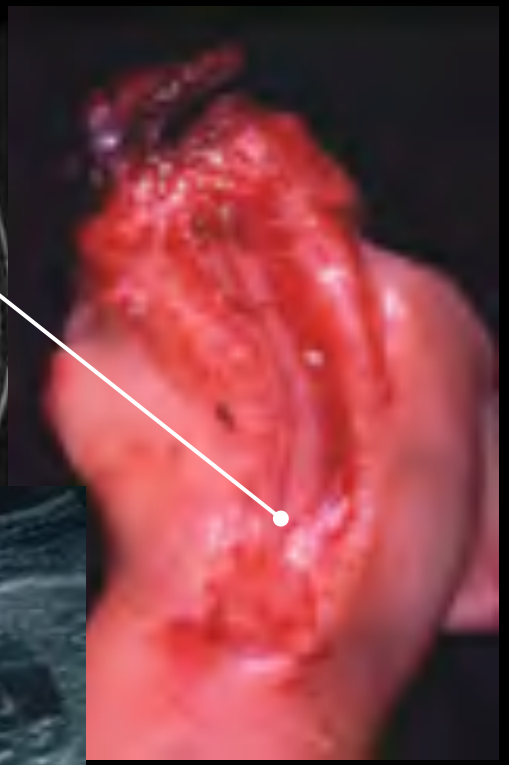
DIFETTI da mancata CHIUSURA o SCHISI del CRANIO

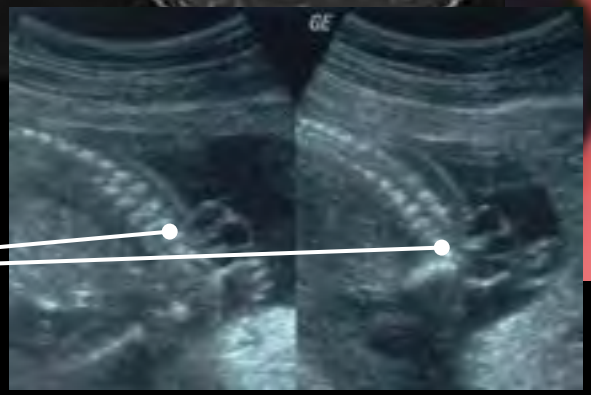
MENINGOENCEFALOCELE

Protrusione delle strutture intracraniche [encefaliche e meningee] attraverso difetti delle ossa del cranio e della dura madre



Erniazione delle strutture cerebellari + meningi della fossa cranica posteriore [e del midollo spinale cervico-toracico]





Diagnosi ecografica prenatale

Am J Med Genet 2005;136:357

Malformazioni telencefaliche

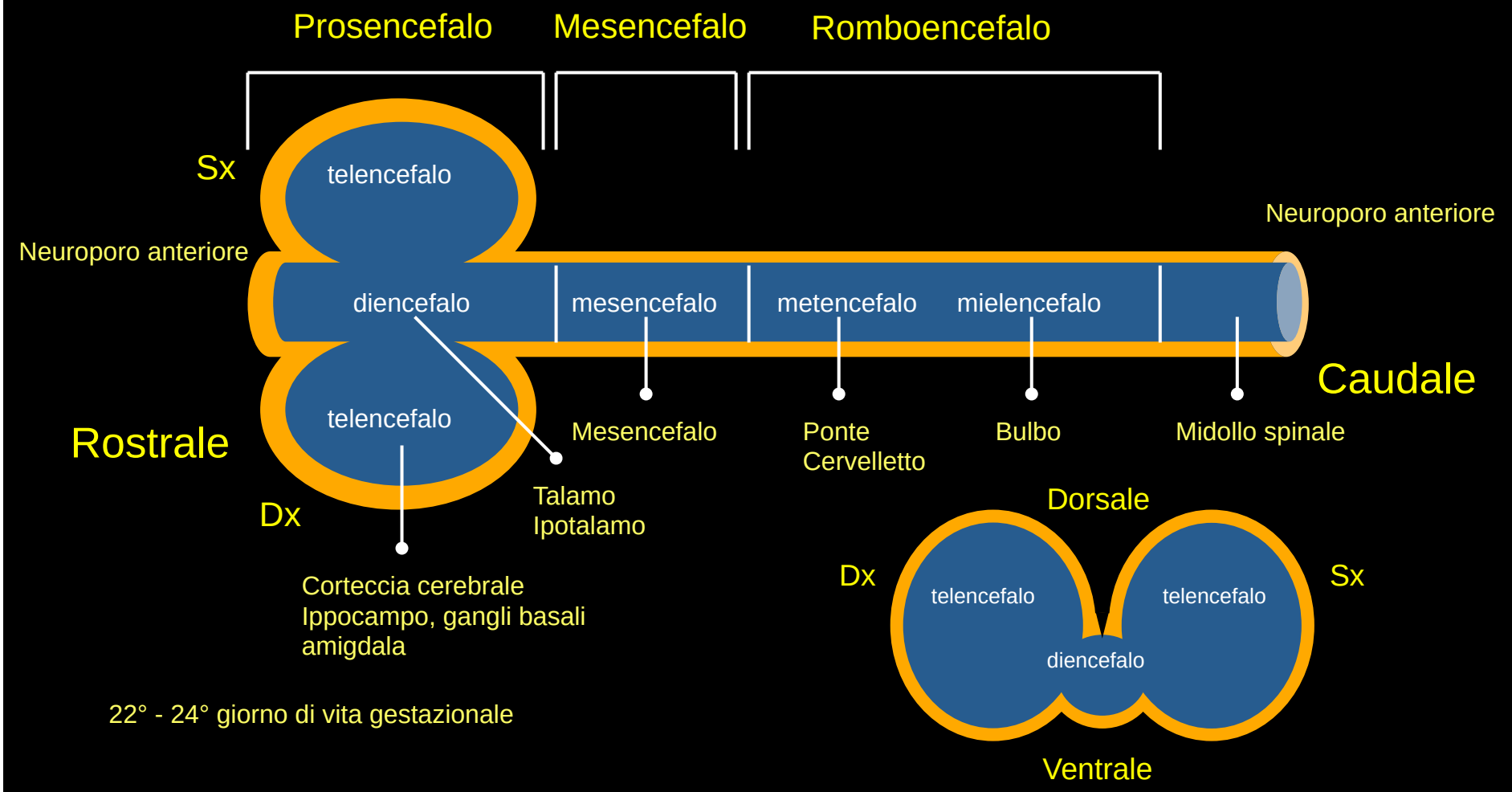
→ MANCA QUALCOSA .....

- *OLOPROSENCEFALIA*
- *Agenesia-ipogenesia CORPO CALLOSO*

→ QUALCOSA VIENE "COSTRUITA" MALE .....

- *MALFORMAZIONI DELLO SVILUPPO CORTICALE*

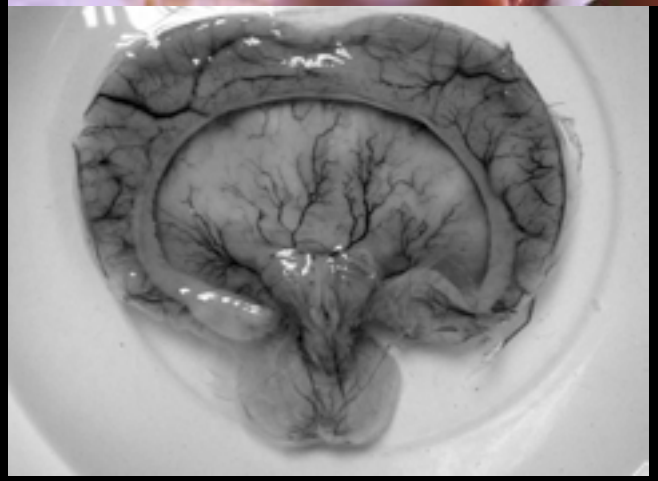
PROSENCEFALO



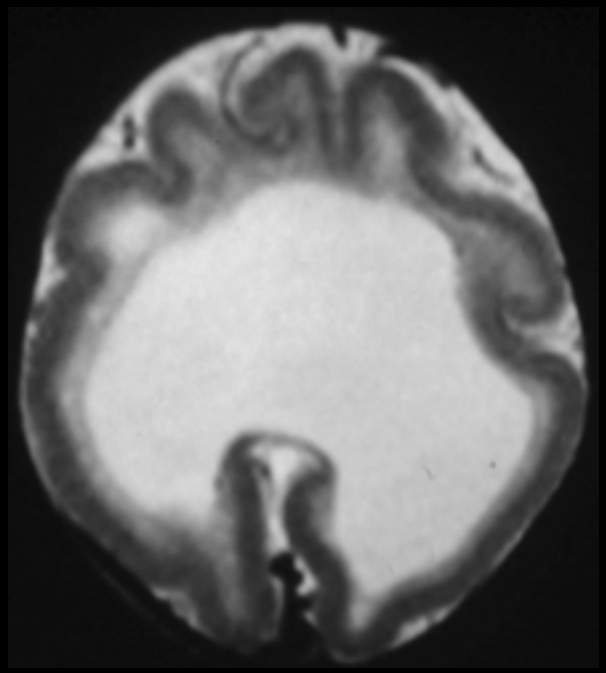
22° - 24° giorno di vita gestazionale

MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

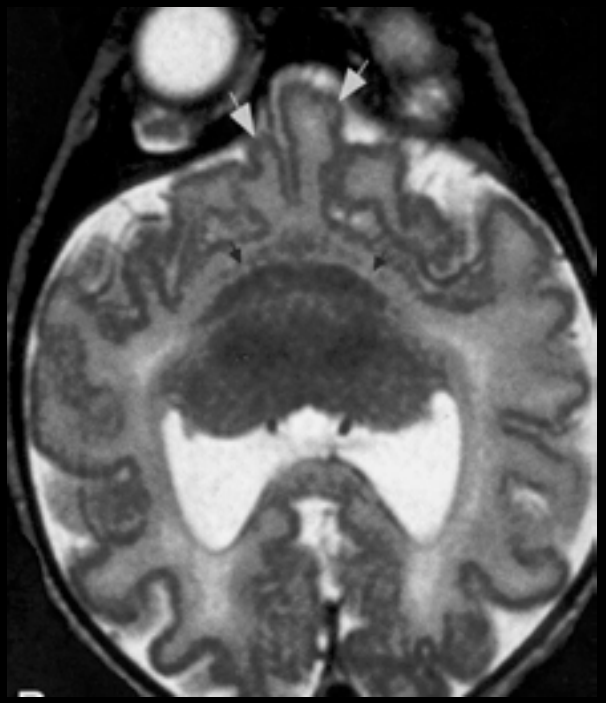
OLOPROSENCEFALIA e MALFORMAZIONI CORRELATE



OLOPROSENCEFALIA



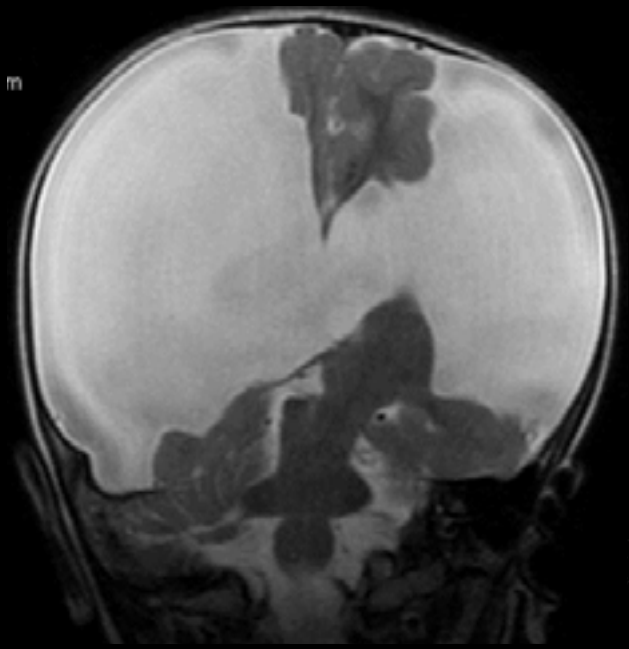
ALOBARE

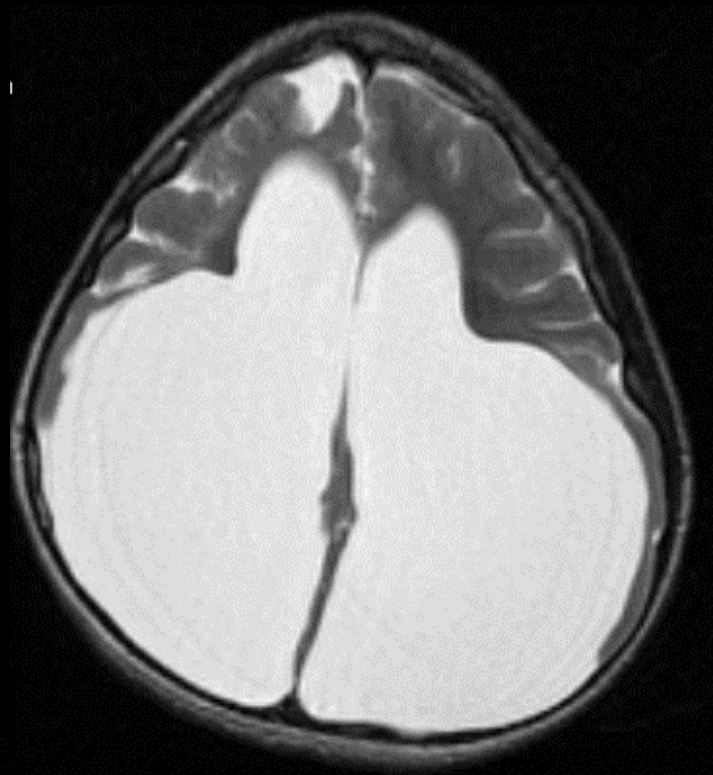


SEMILOBARE

LOBARE

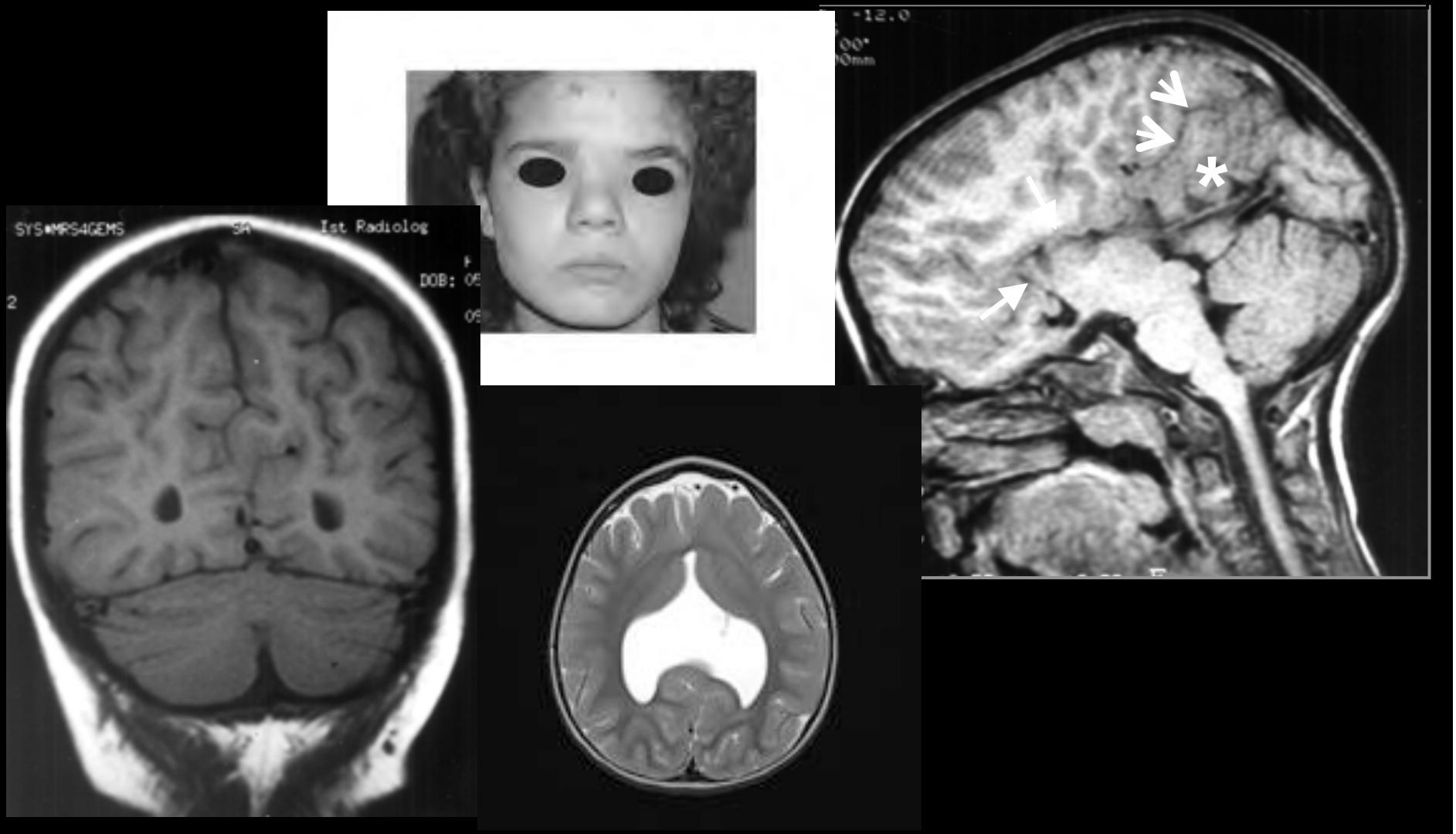
Assenza del setto pellucido





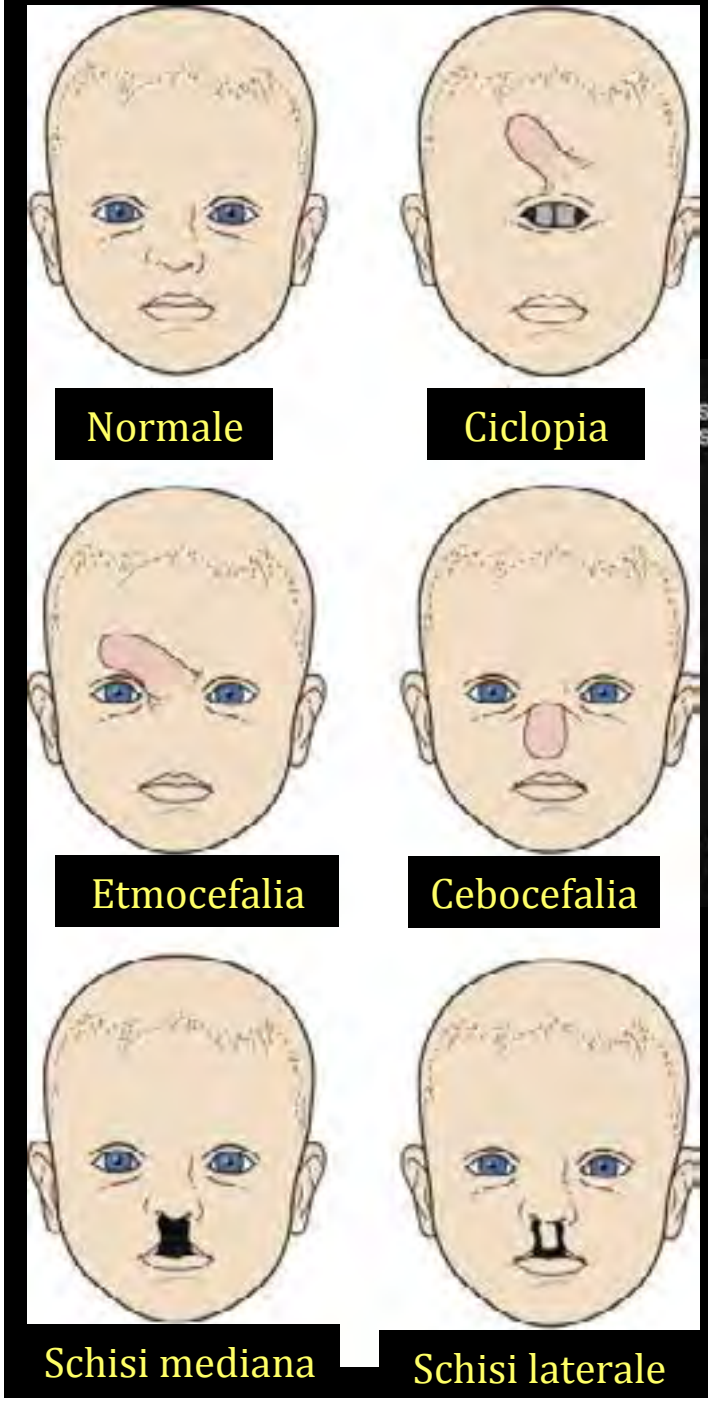
MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

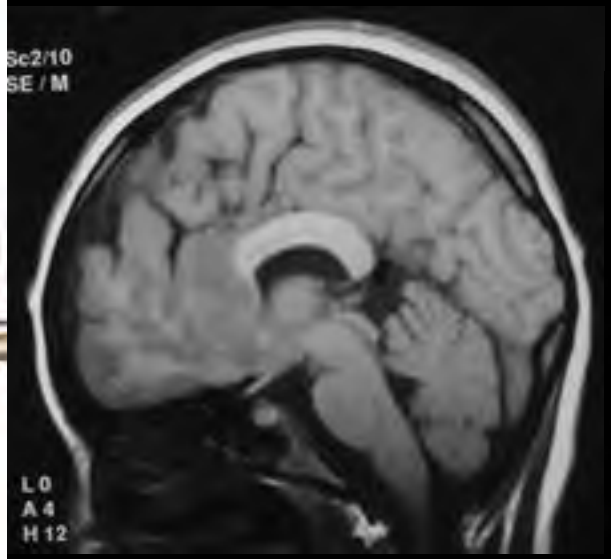
OLOPROSENCEFALIA e MALFORMAZIONI CORRELATE



MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

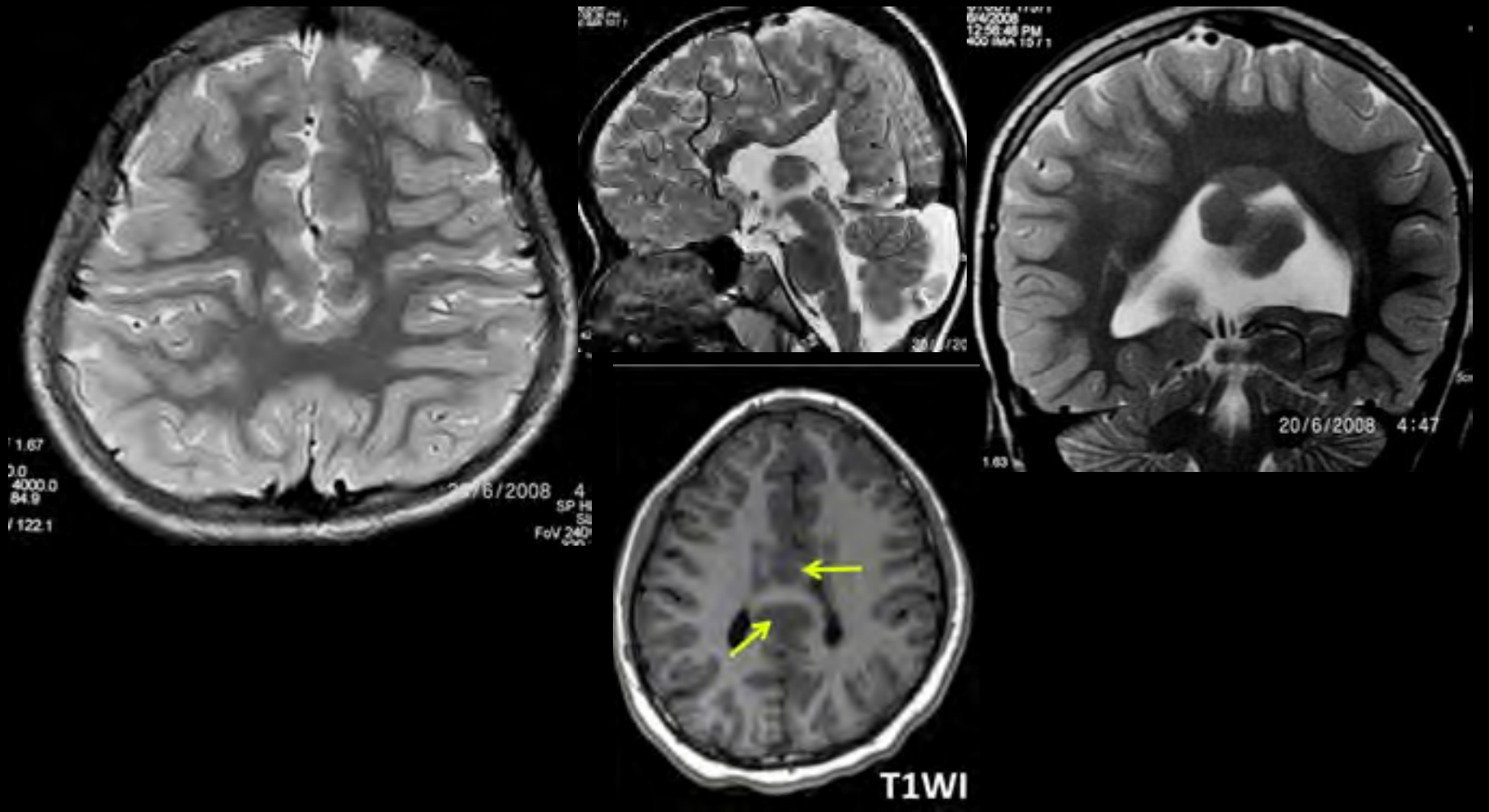
OLOPROSENCEFALIA e MALFORMAZIONI CORRELATE





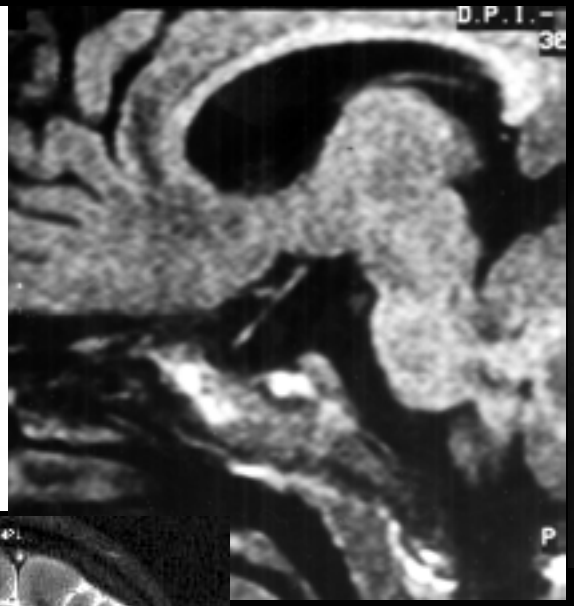
MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

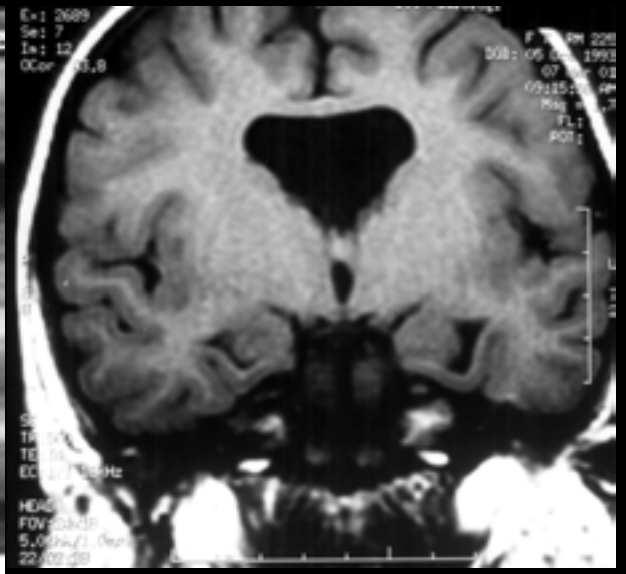
SINTELENCEFALIA

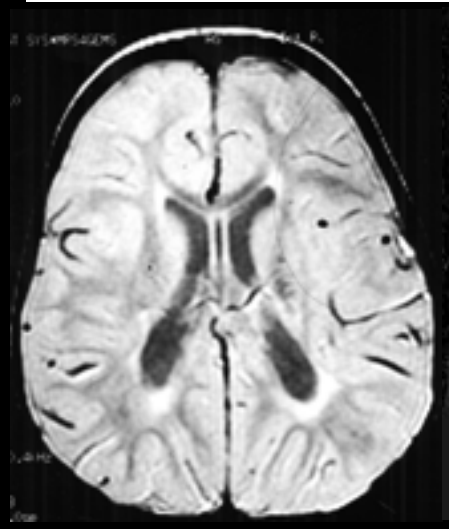


MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

DISPLASIA SETTO-OTTICA

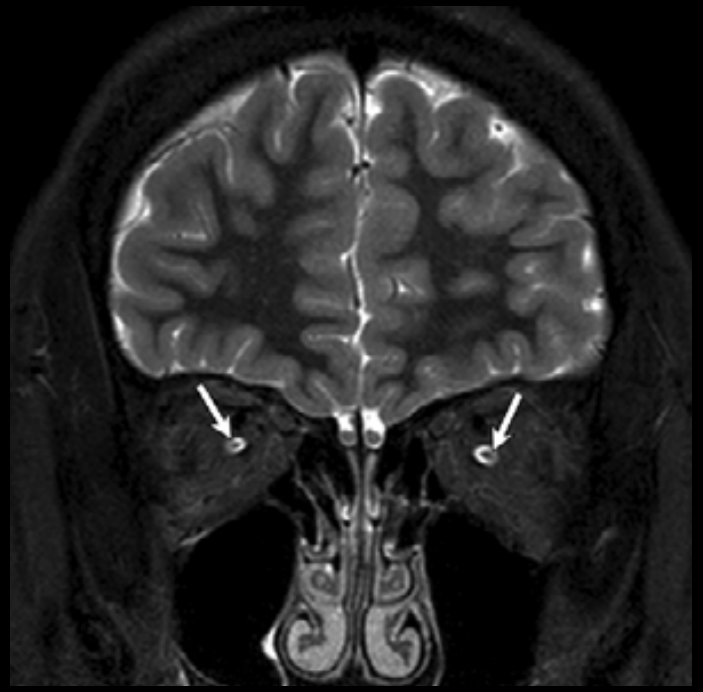


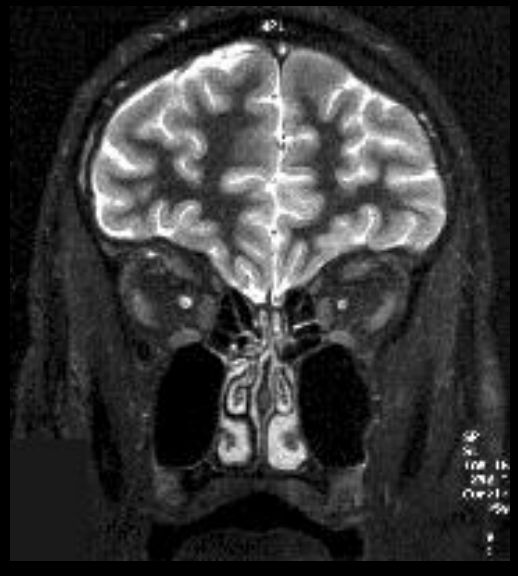




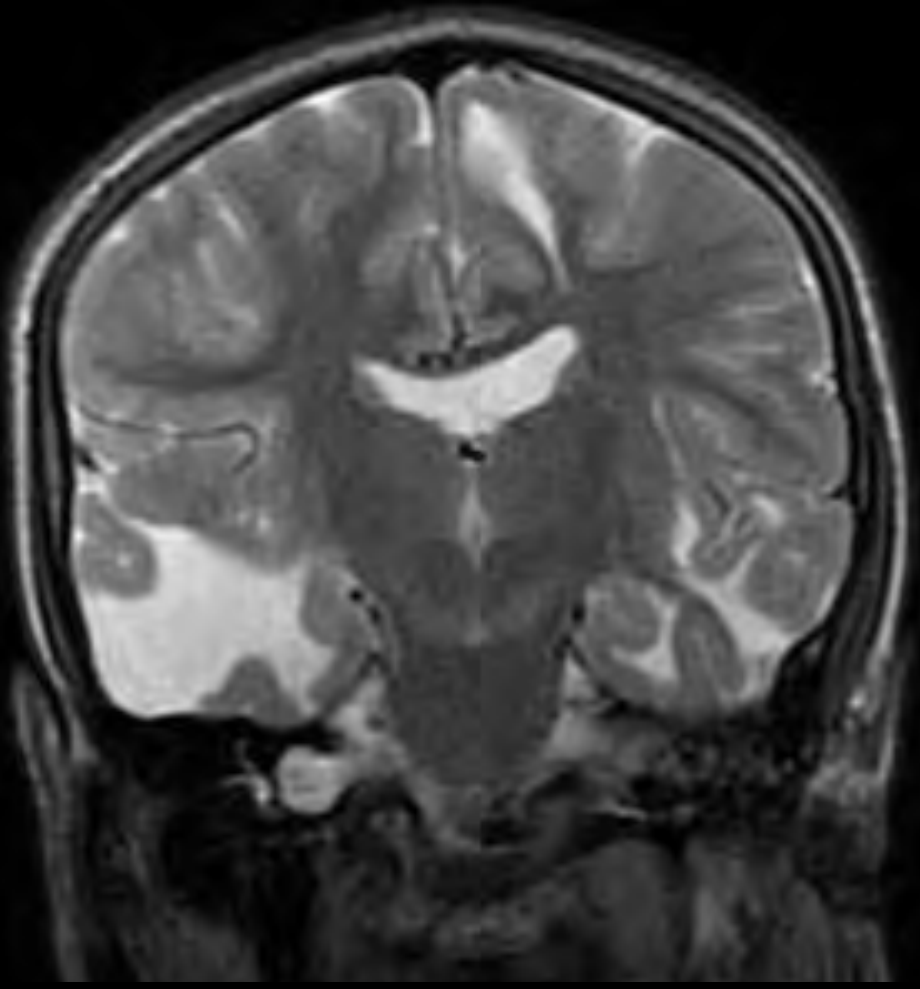
MALFORMAZIONI del PROSENCEFALO MEDIOBASALE

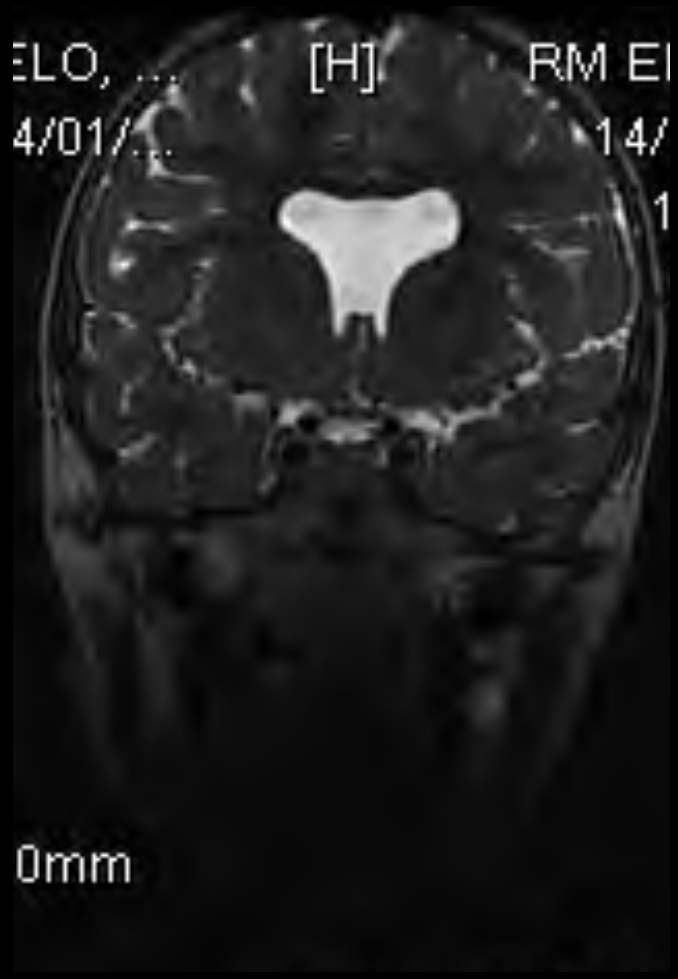
DISPLASIA SETTO-OTTICA



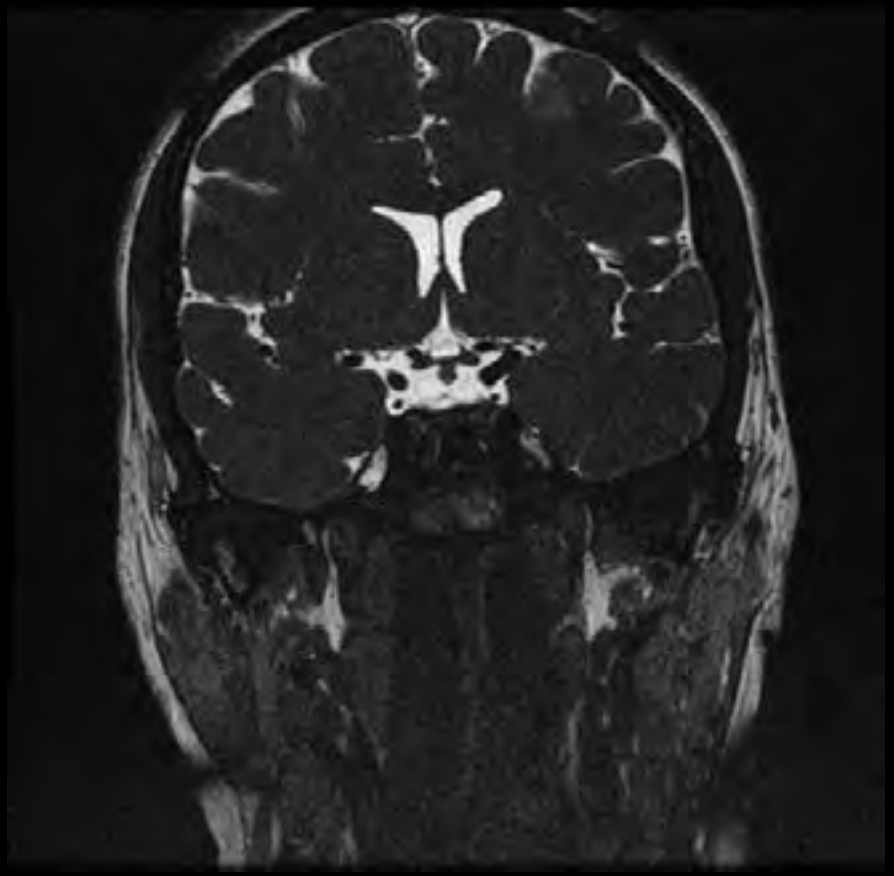


Agenesia del setto pellucido con malformazioni associate





Pan ipopituitarismo



Corpo calloso

Lamina rostrale e Rostro

Ginocchio

Corpo [tronco]

Istmo

Splenio

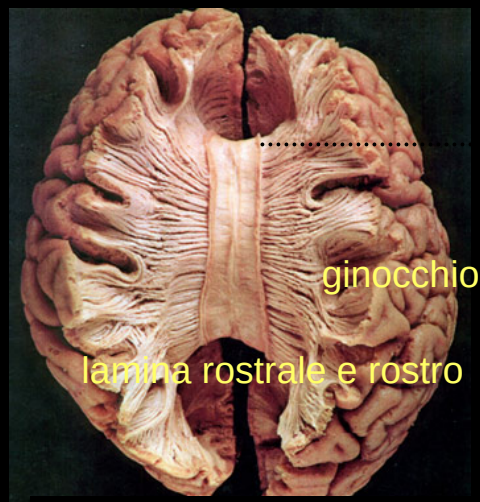
Commissure [forcipi] principali & accessori

MALFORMAZIONI delle COMMISSURE CEREBRALI

COMMISSURE TELENCEFALICHE

- Commissura anteriore --> fibre profonde [3.5 milioni]
- Commissura ippocampale --> “psalterio”
- Corpo calloso --> fibre omotopiche; non reciproche; eterotopiche [200 milioni]

CORPO CALLOSO [connette la neocorteccia dei due emisferi]



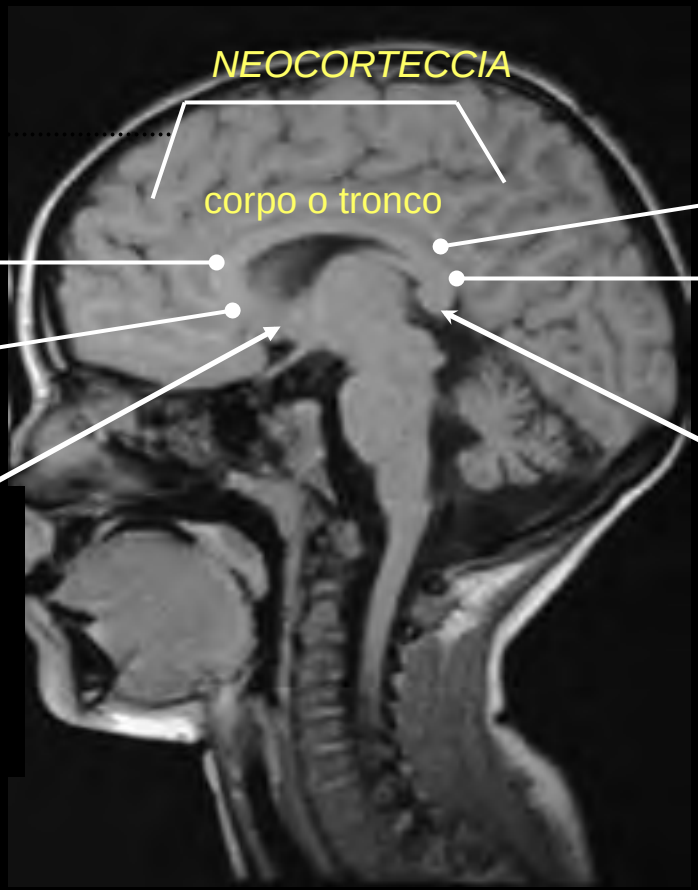
ginocchio

lamina rostrale e rostro

Commissura ANTERIORE

PALEOCORTECCIA

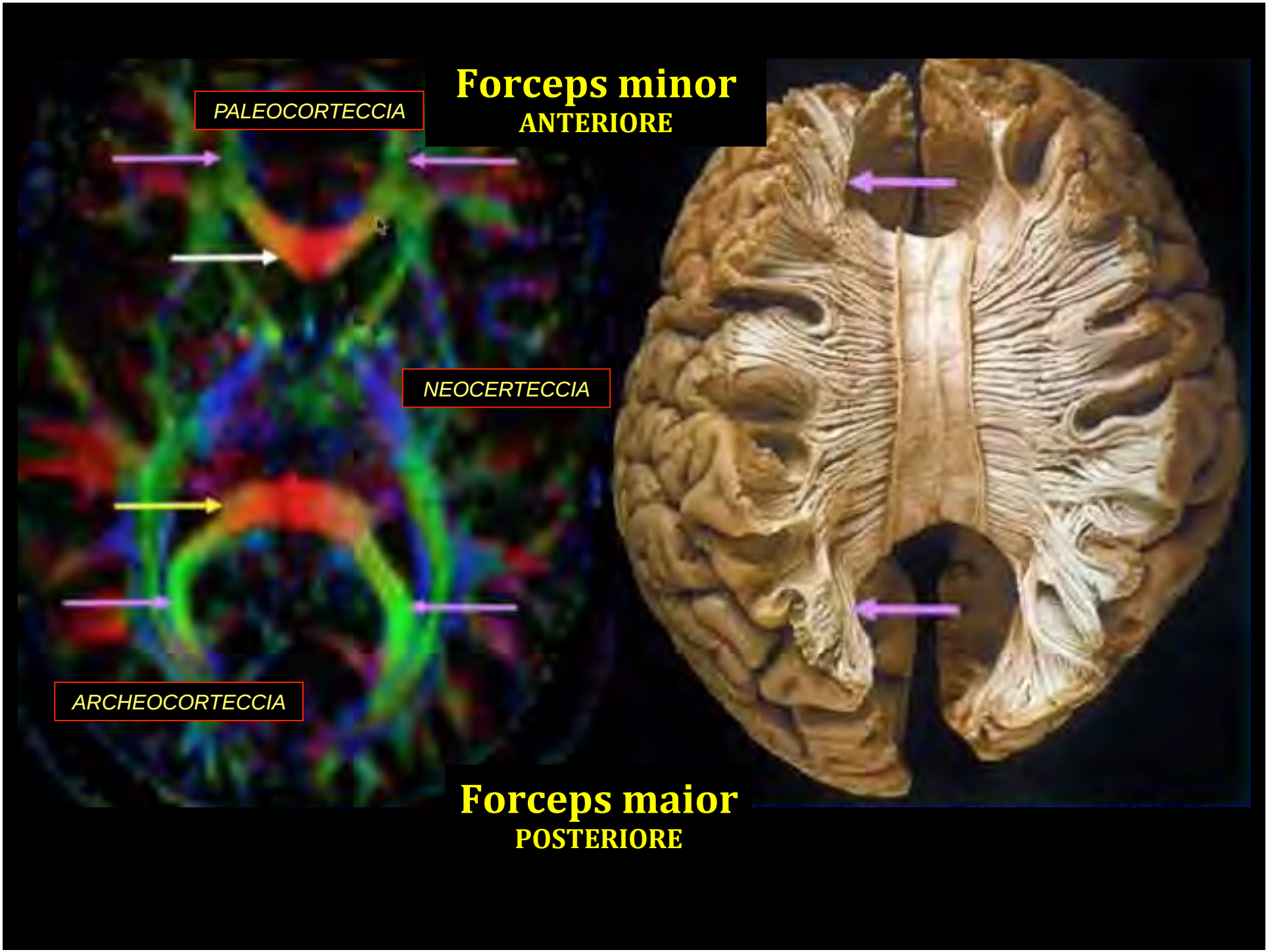
[connette la corteccia olfattiva dei due emisferi]



Commissura IPPOCAMPALE

ARCHEOCORTECCIA

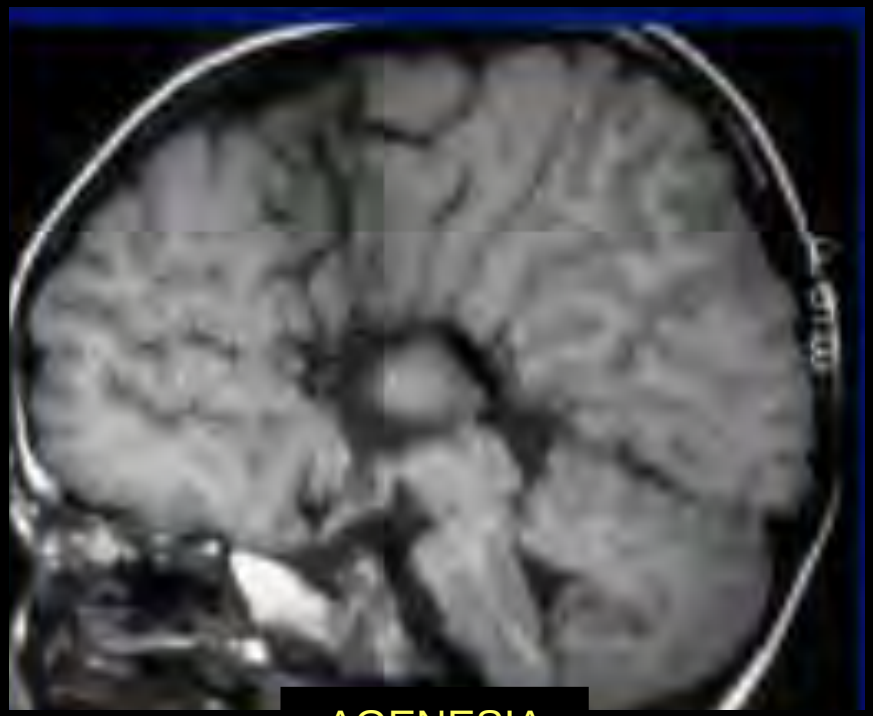
[connette la corteccia del rinencefalo --> sistema limbico]


Forceps minor
ANTERIORE
PALEOCORTECCIA
NEOCERTECCIA
ARCHEOCORTECCIA
Forceps maior
POSTERIORE

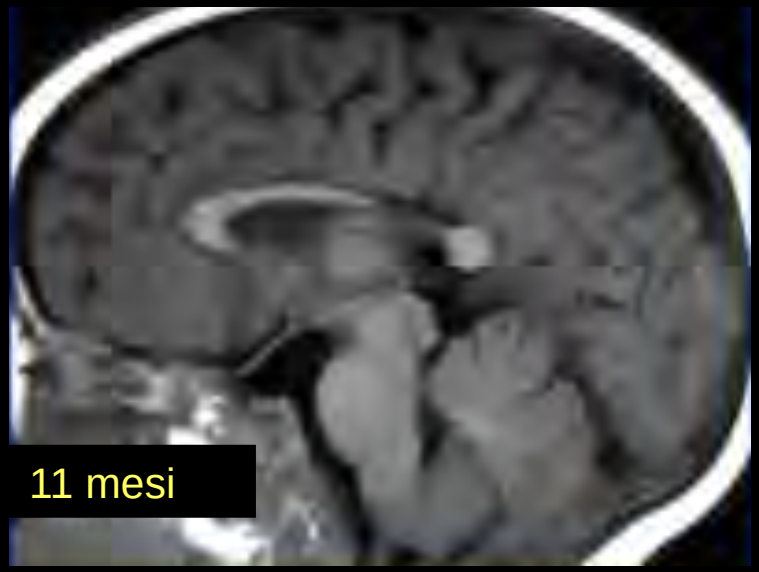
AZZURRO → cranio – caudale e viceversa

ROSSO → da destra a sinistra e viceversa

VERDE → antero – posteriore e viceversa



AGENESIA



DISGENESIA?
Distruzione segmentale?

….. la conoscenza della normale sequenza delle tappe dello sviluppo aiuta a distinguere tra DISGENESIA e lesioni DISTRUTTIVE secondarie

……

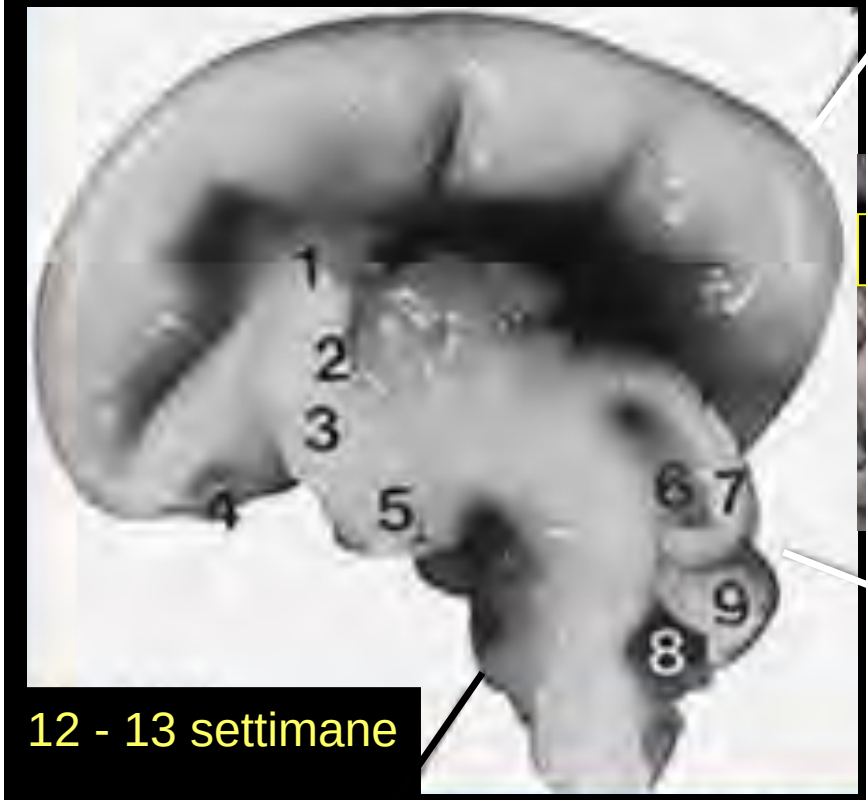
Quale è la parte del CC che appare per prima ?

"VECCHIA SEQUENZA" → Ginocchio→ Corpo → Splenio → Rostro

→ NON ha senso da un punto di vista evoluzionistico ……..

Se il Ginocchio fosse davvero la PRIMA struttura a svilupparsi, il CORPO del corpo calloso non potrebbe essere presente in assenza di ginocchio……..

SVILUPPO delle COMMISSURE



12 - 13 settimane

Commissura ANTERIORE → 9 - 10 settimane

Commissura IPPOCAMPO → 10 - 11 settimane

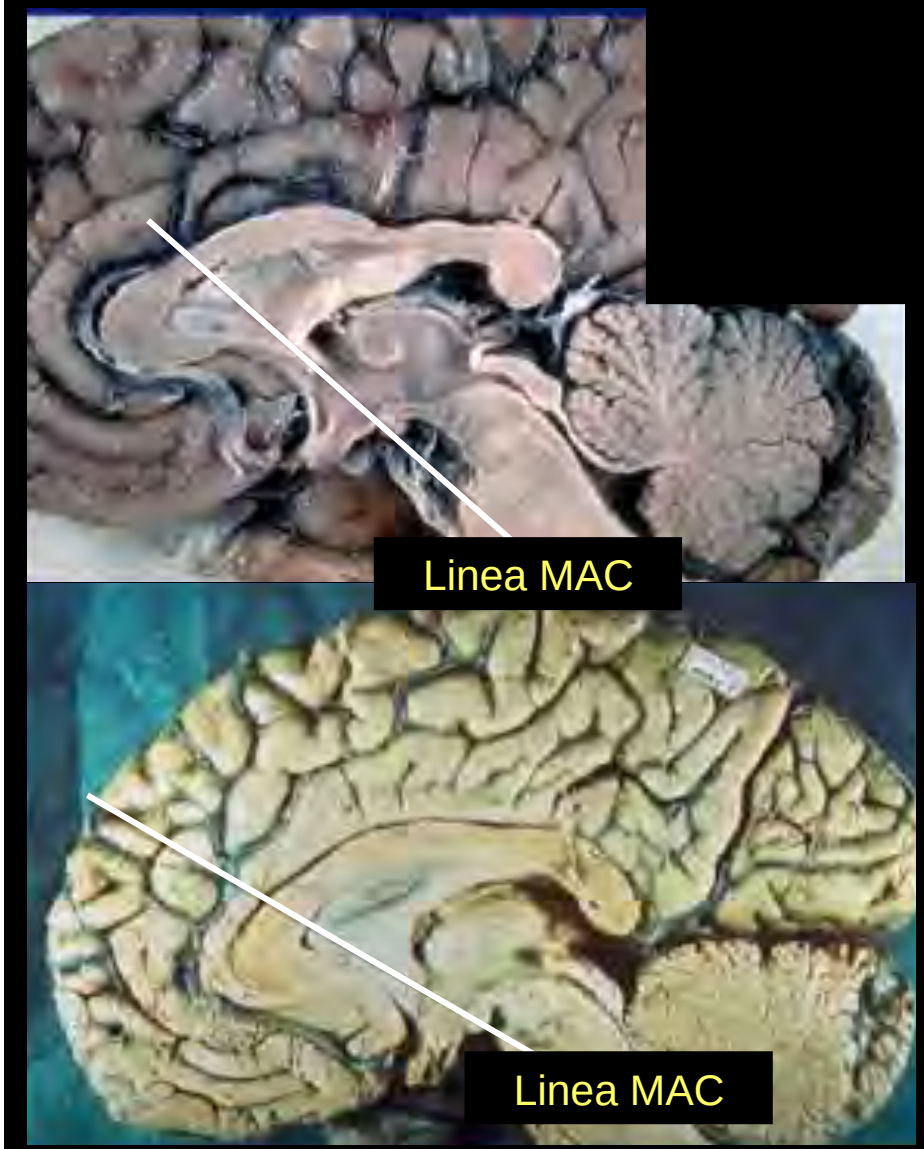
CORPO CALLOSO → 11 - 12 settimane

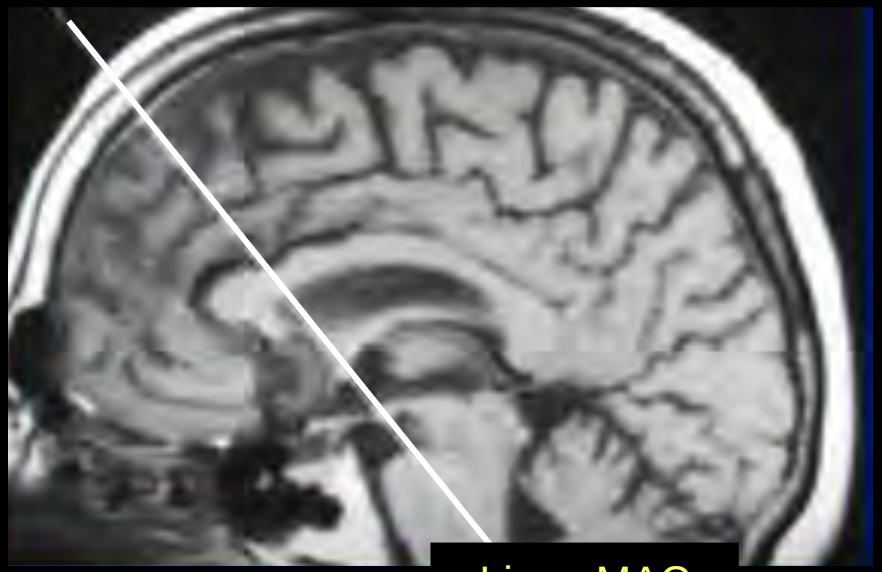
La “*linea* MAC” [GINOCCHIO → giunzione anteriore CORPO]

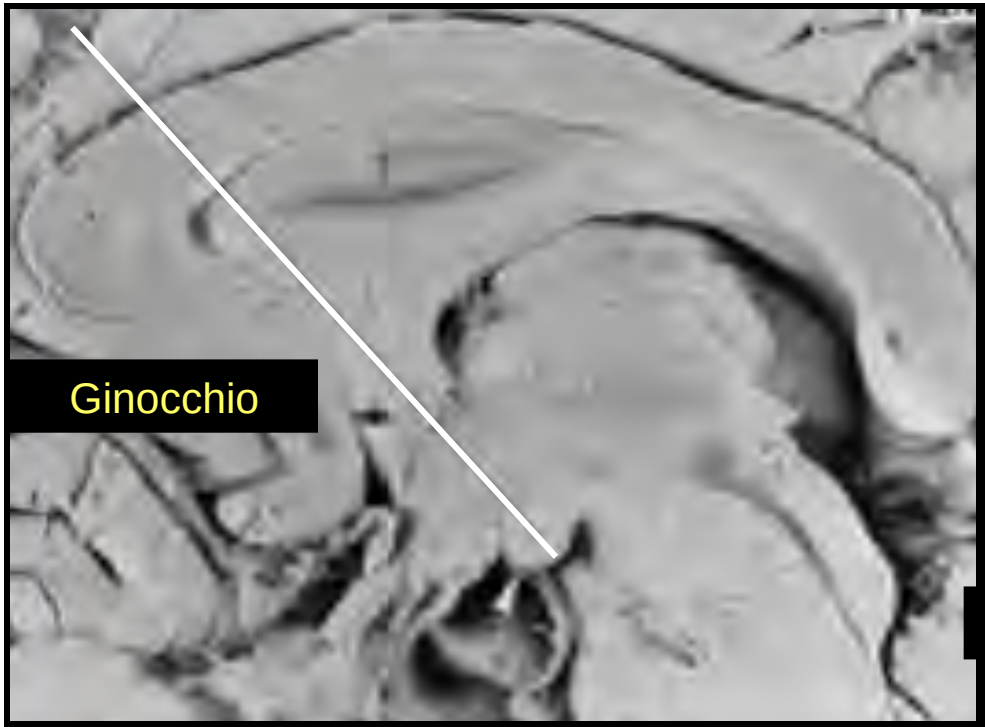
Corpi MAMMILLARI

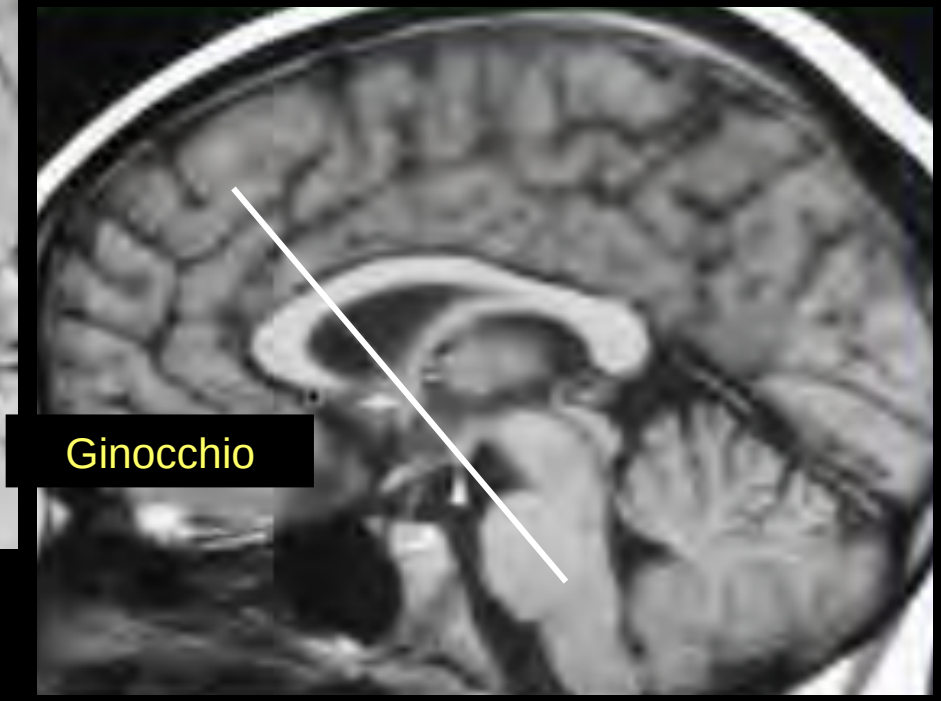
Commissura ANTERIORE

CORPO CALLOSO [ginocchio]

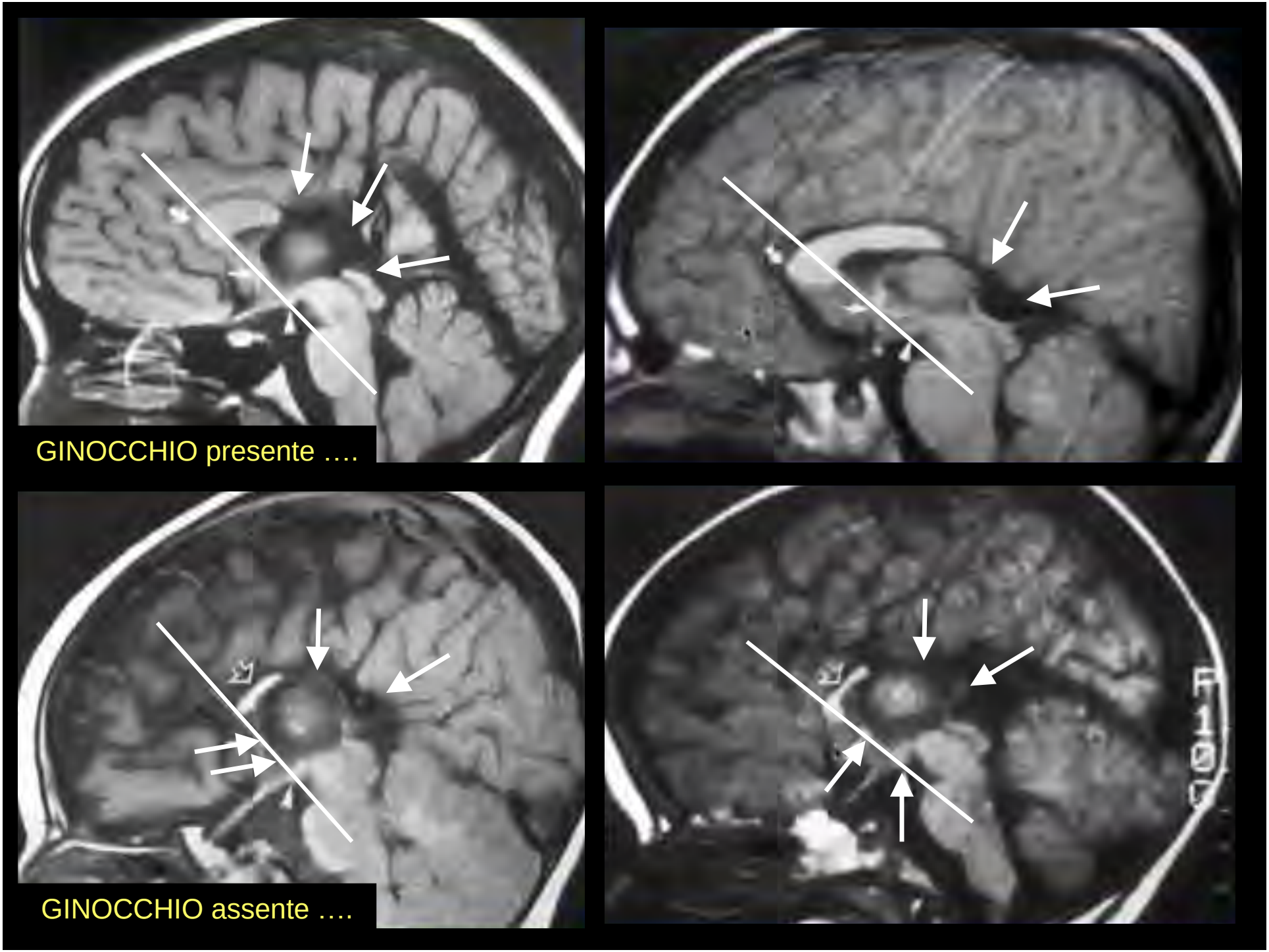


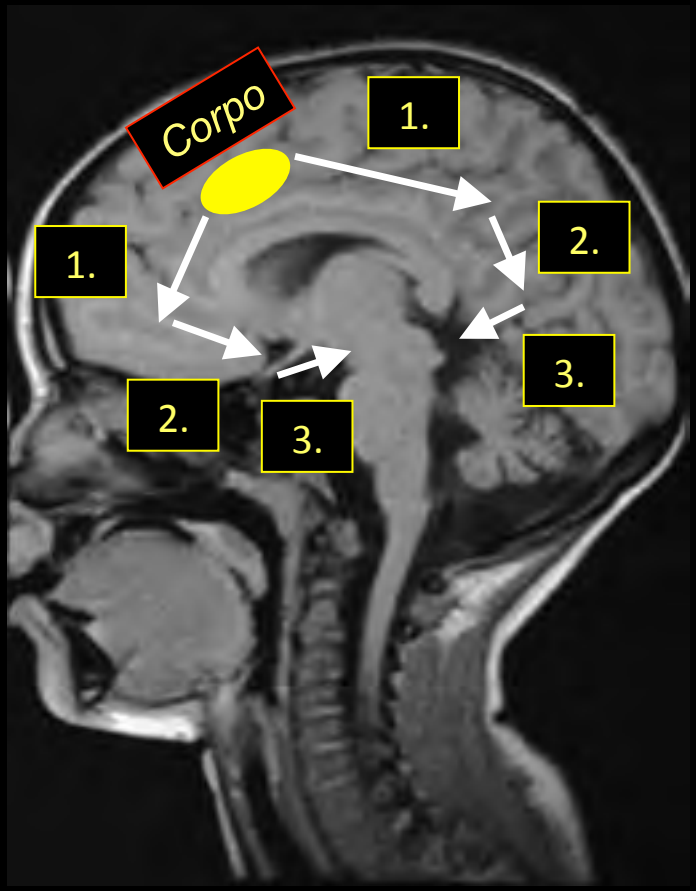






…. il GINOCCHIO normale è sempre localizzato di fronte
alla linea MAC ……


GINOCCHIO presente ….
GINOCCHIO assente ….

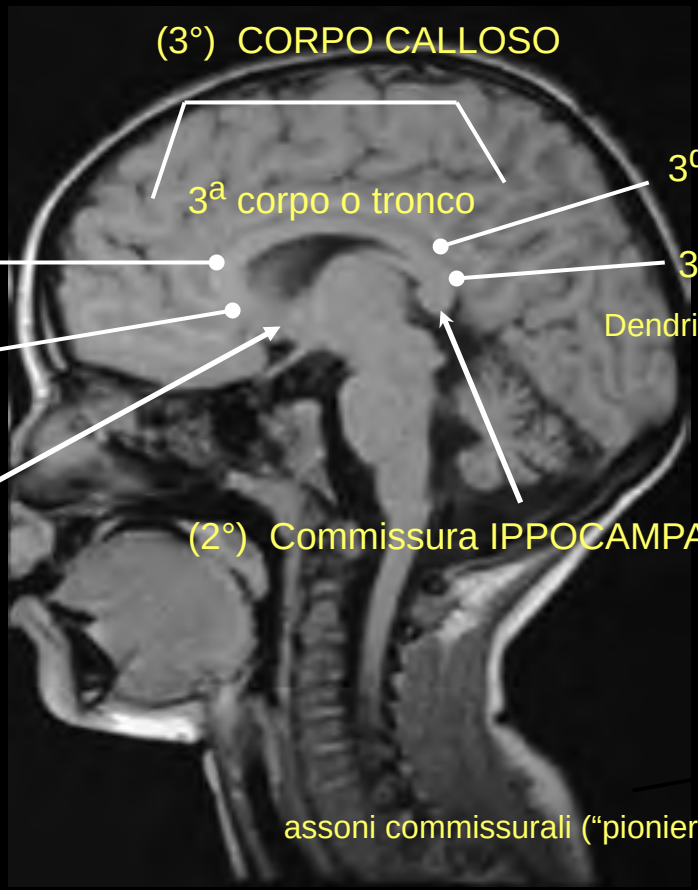


……. il CORPO CALOSO inizia a svilupparsi dalla regione anteriore del corpo e cresce bi-direzionalmente …..

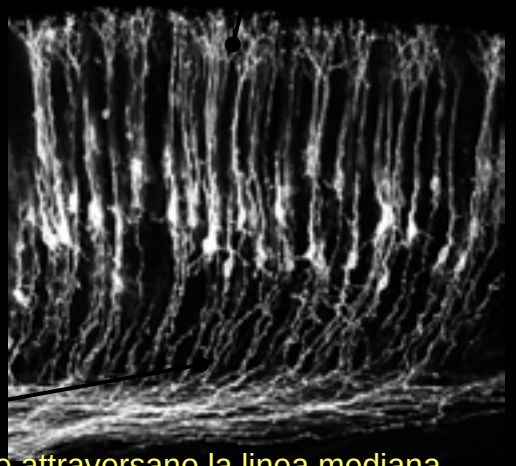
MALFORMAZIONI delle COMMISSURE CEREBRALI

COMMISSURE TELENCEFALICHE --> TAPPE di sviluppo embriologico

[a - 20a settimana di vita intrauterina] --> malformazioni cerebrali associate



Dendriti neuronali dell'emisfero controlaterale

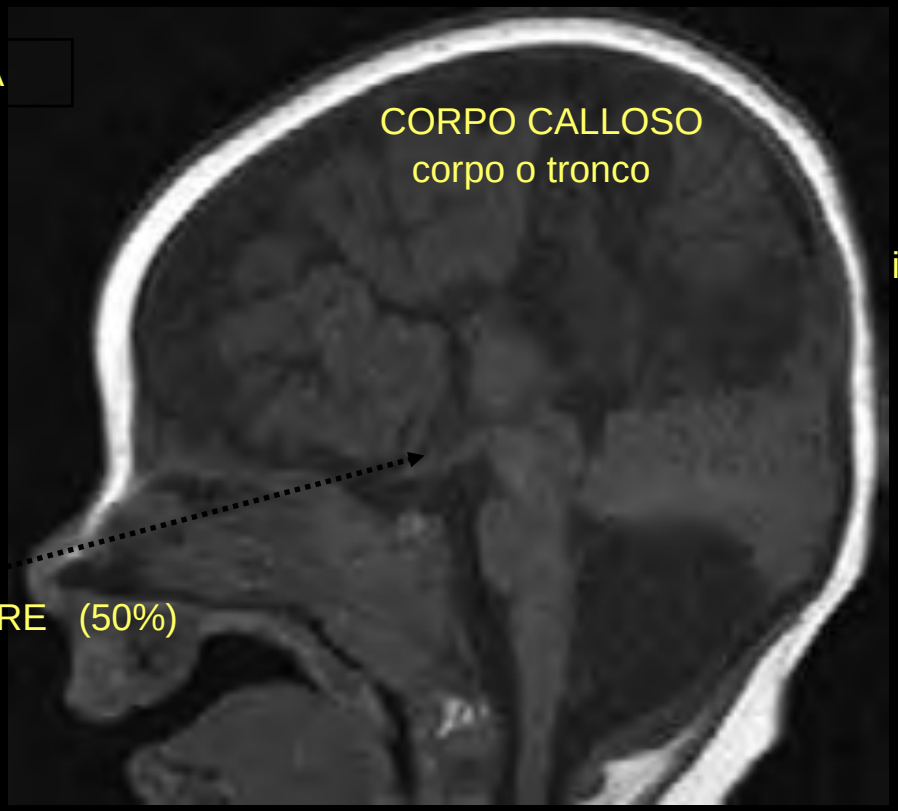


assoni commissurali ("pionieri") che attraversano la linea mediana

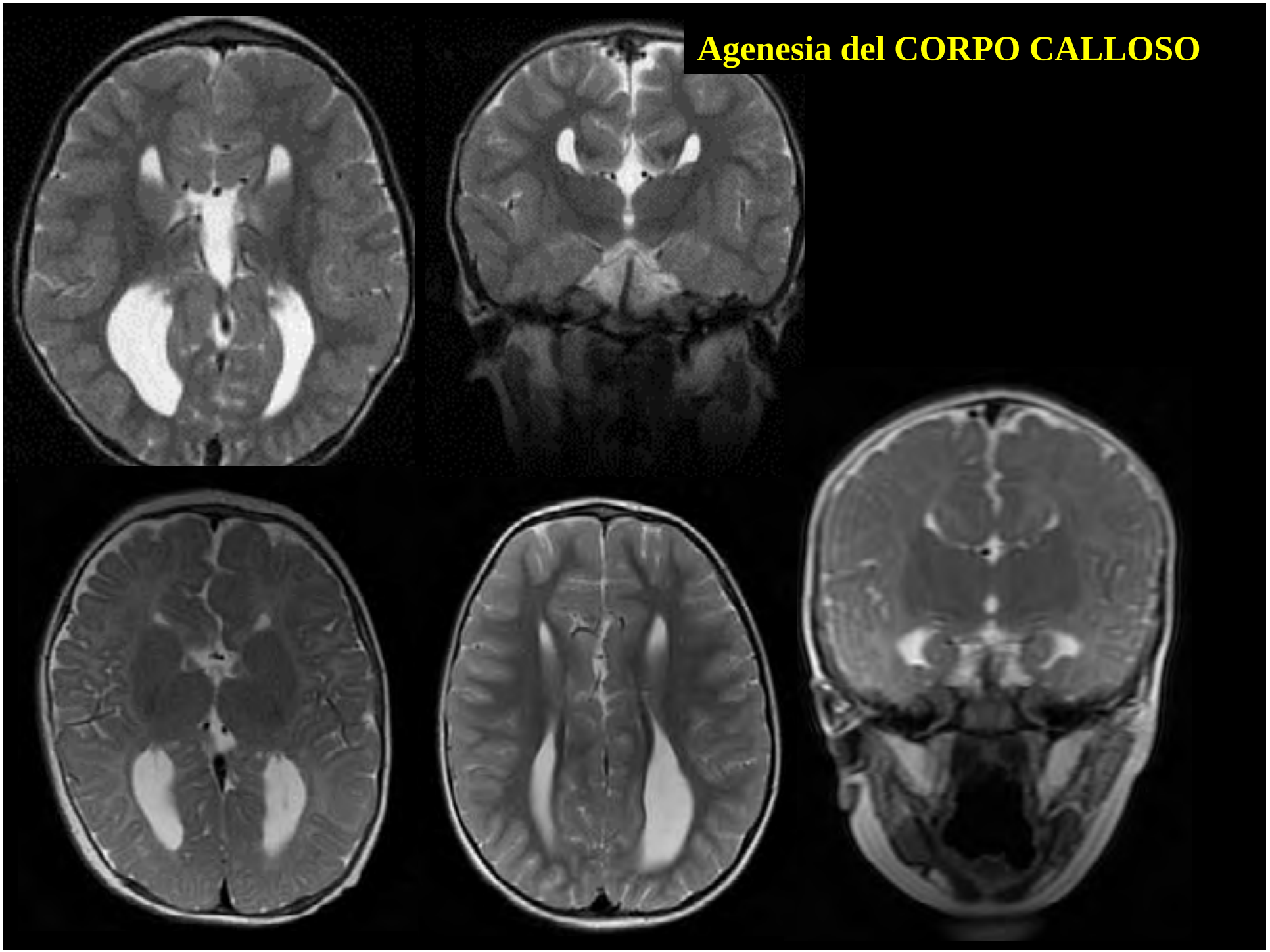
MALFORMAZIONI delle COMMISSURE CEREBRALI

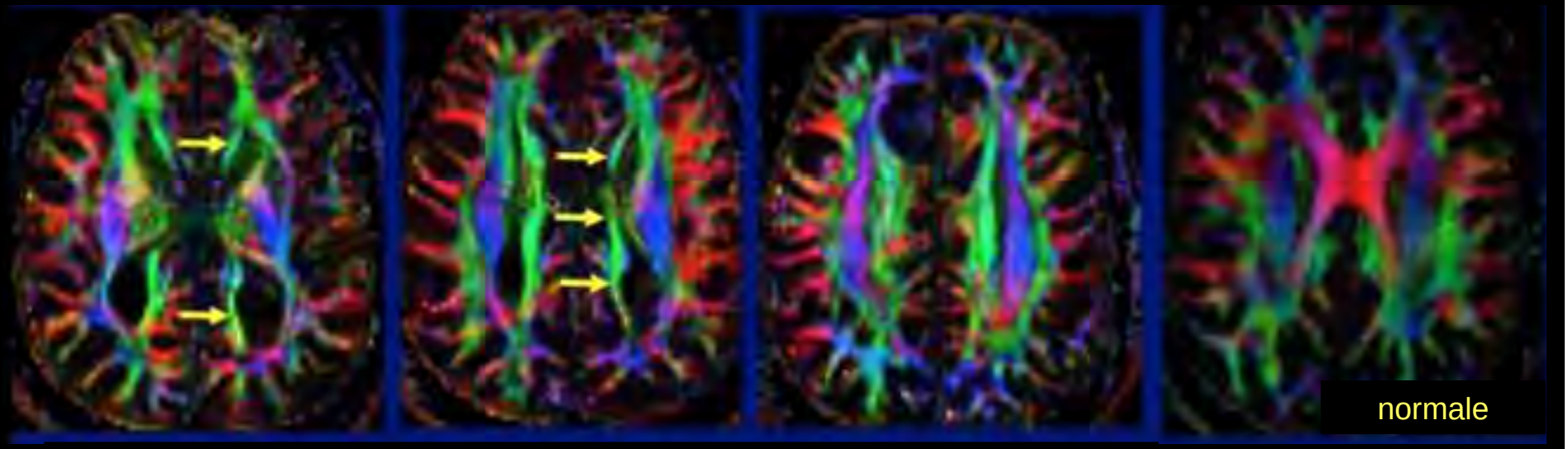
•AGENESIA COMPLETA dI tutte le COMMISSURE TELENCEFALICHE

FORMA COMPLETA



Acta Paediatr 2005;94:1066-1072


Agenesia del CORPO CALLOSO



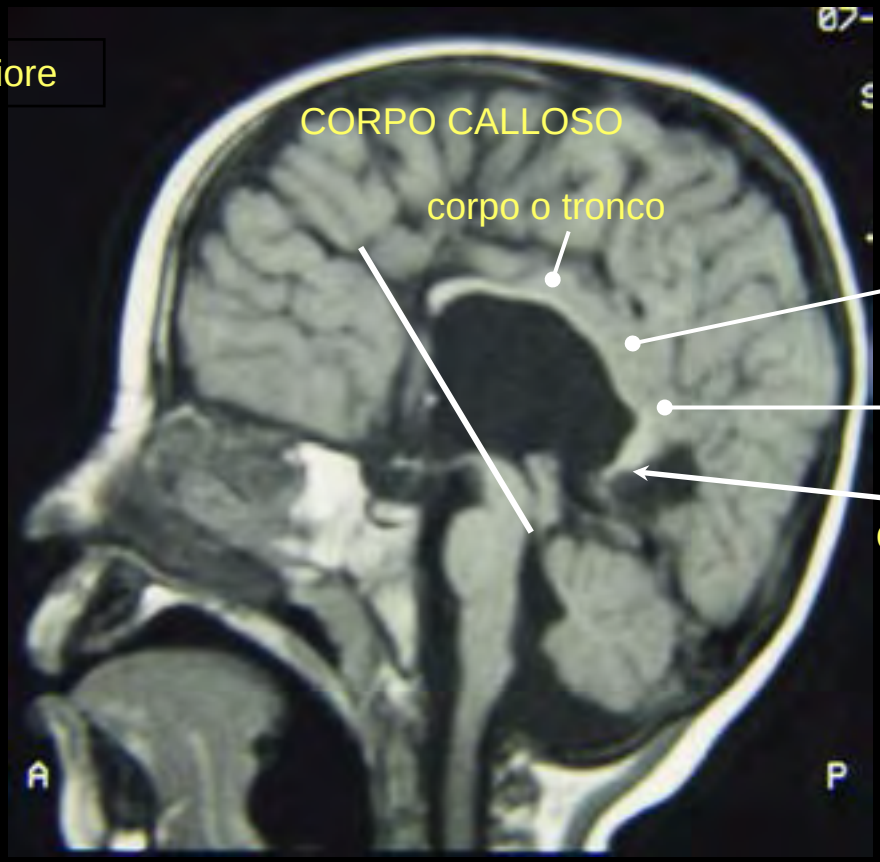
Fascicoli longitudinali del corpo calloso [fasci di Probst]

Agenesia del corpo calloso

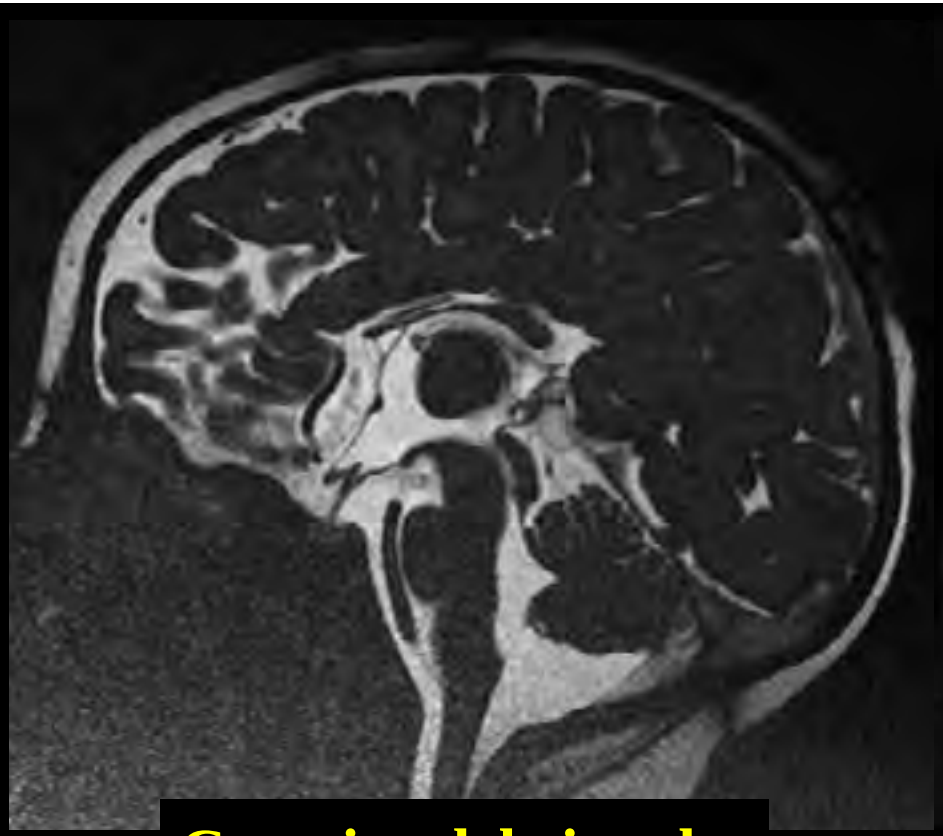
MALFORMAZIONI delle COMMISSURE CEREBRALI

AGENESIA PARZIALE delle COMMISSURE TELENCEFALICHE

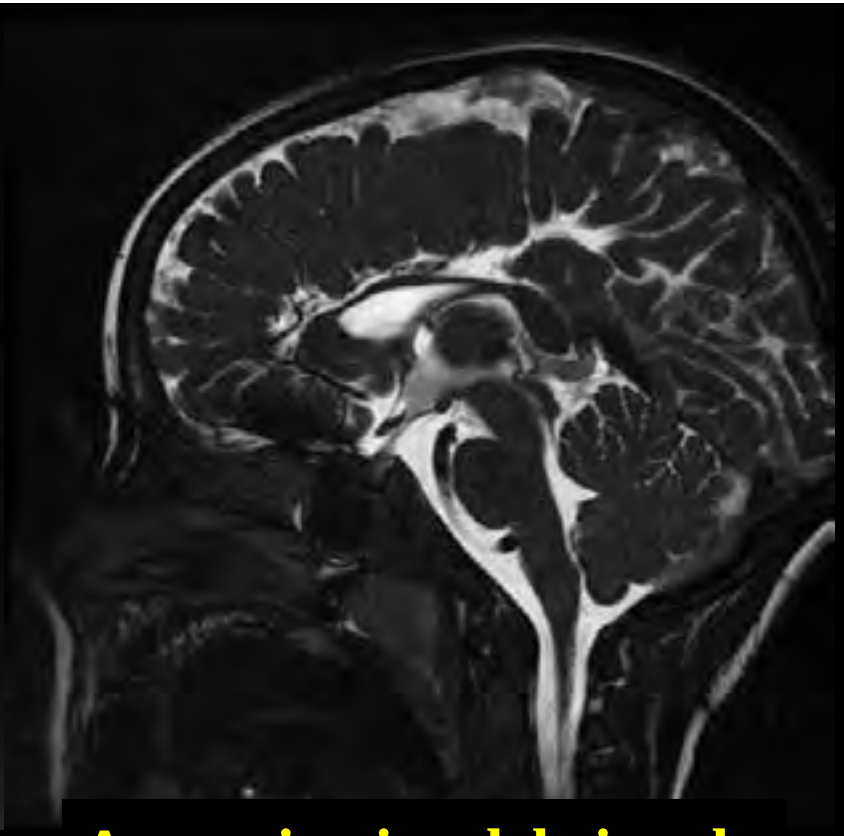
FORMA PARZIALE anteriore



Acta Paediatr 2005;94:1066-1072



Con giro del cingolo



Agenesia giro del cingolo

MALFORMAZIONI delle COMMISSURE CEREBRALI

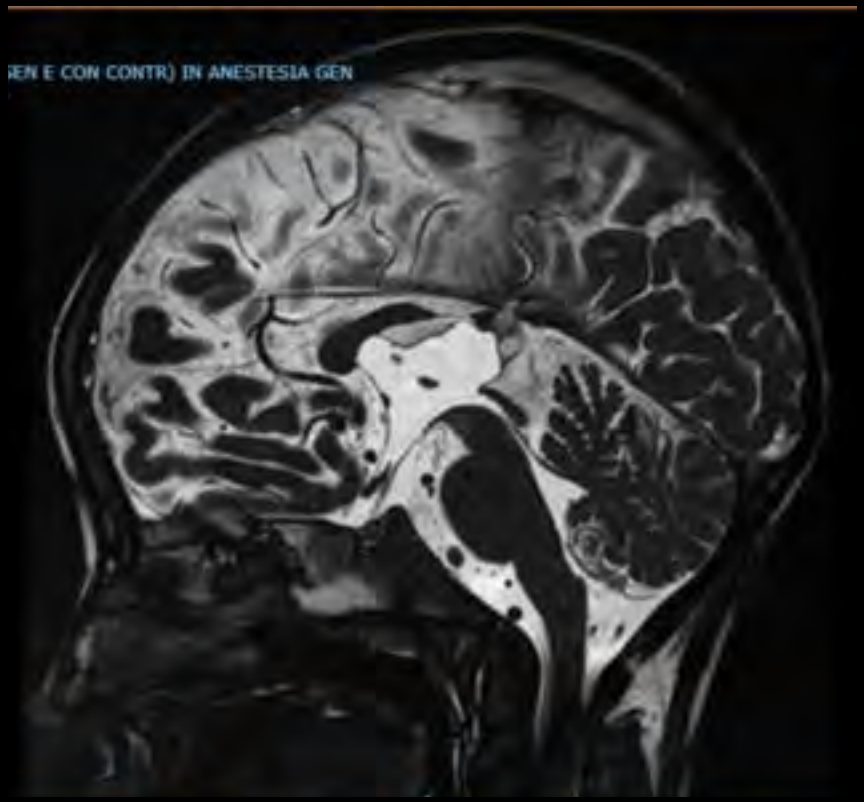
AGENESIA PARZIALE delle COMMISSURE TELENCEFALICHE

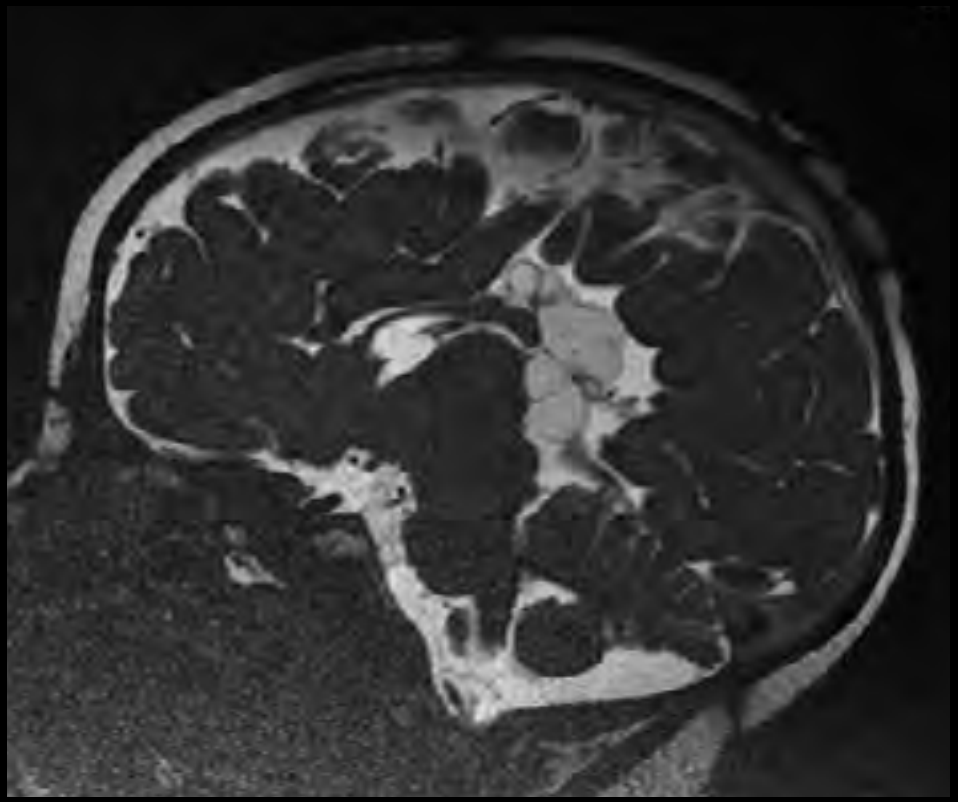
AGENESIA ISOLATA della COMMISSURA IPPOCAMPALE

Acta Paediatr 2005;94:1066-1072

•MALFORMAZIONI delle COMMISSURE CEREBRALI

AGENESIA PARZIALE delle COMMISSURE TELENCEFALICHE

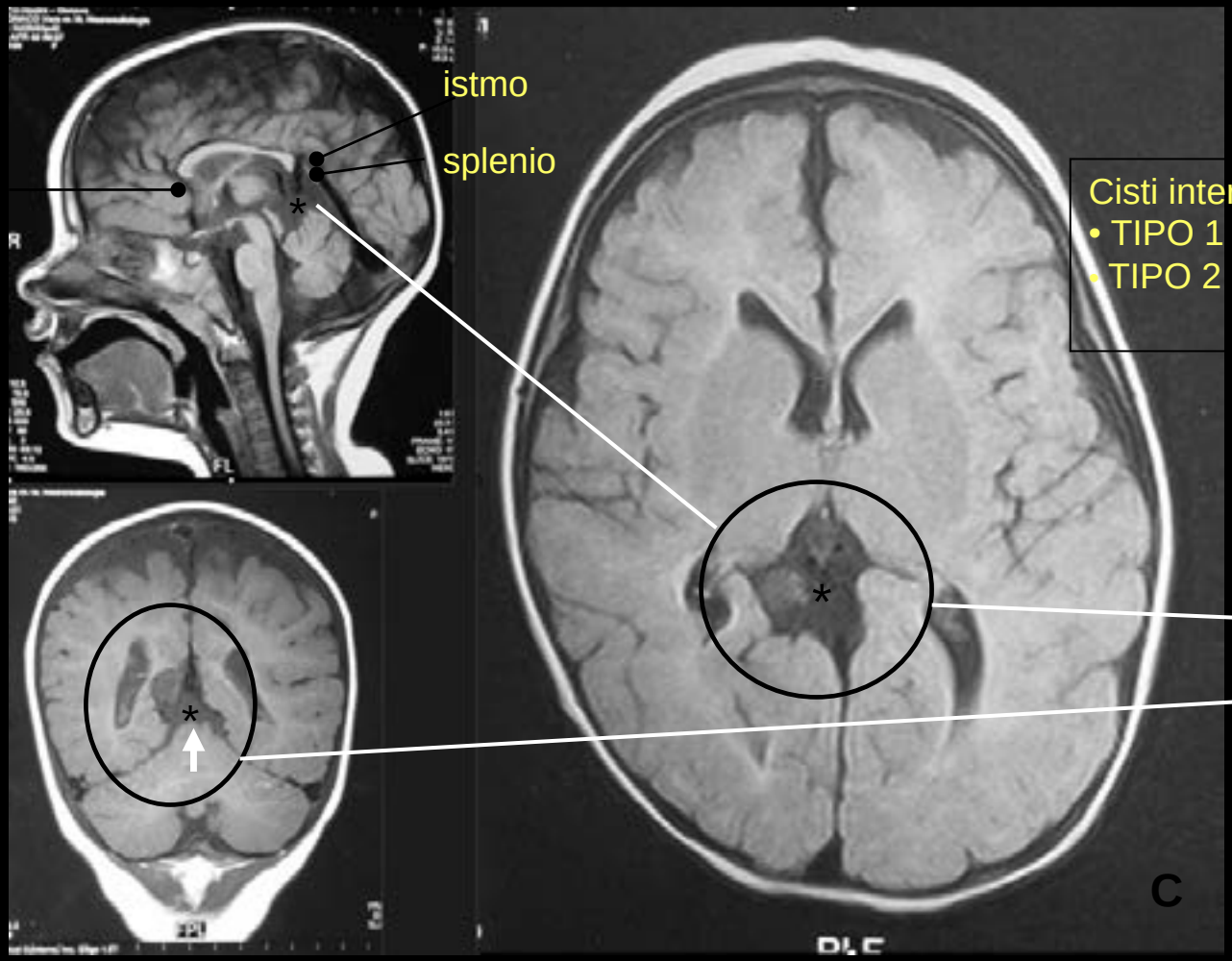




MALFORMAZIONI delle COMMISSURE CEREBRALI

AGENESIA delle COMMISSURE con DISPLASIA delle MENINGI

Agenesia del CORPO CALLOSO con displasia MULTICISTICA delle meningi interemisferiche

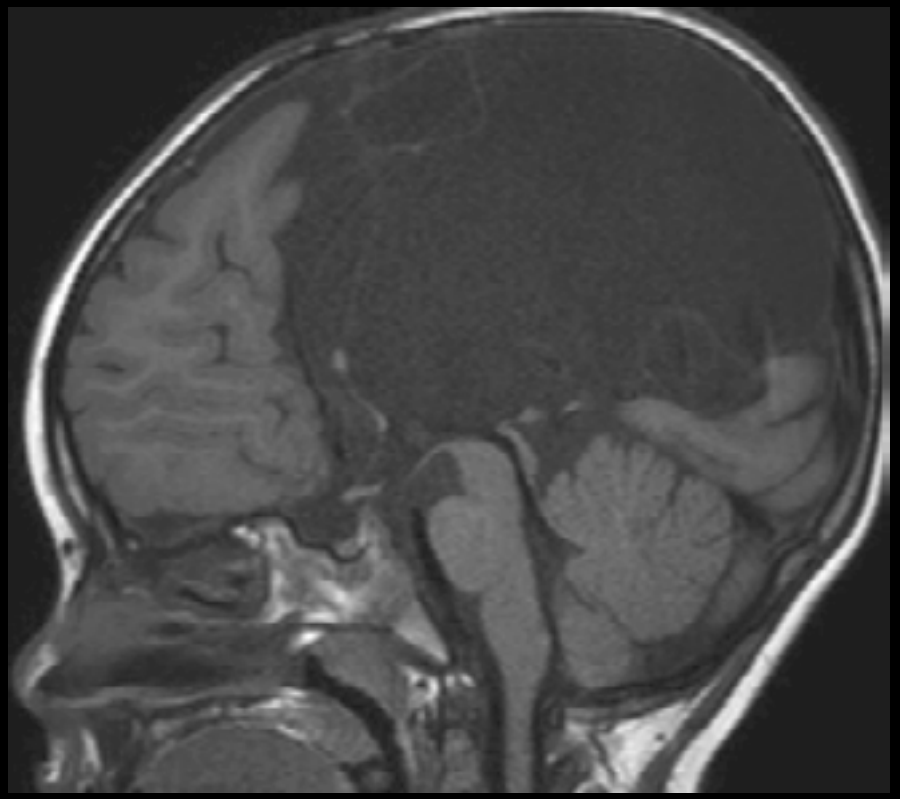


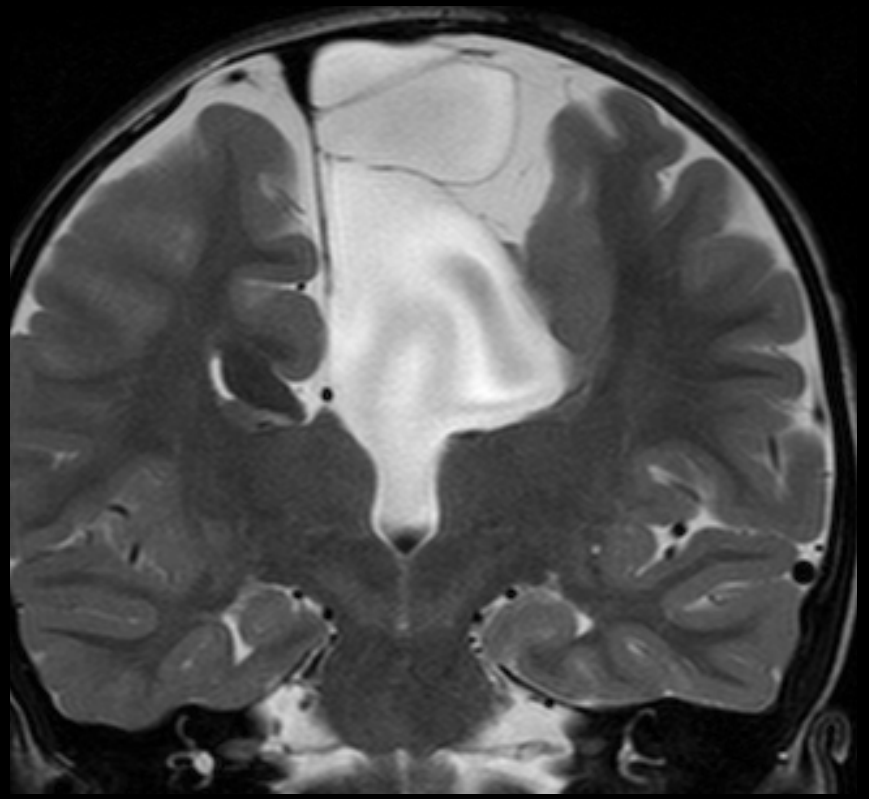
Cisti interemisferica
- TIPO 1 --> comunicante con ventricoli
- TIPO 2 --> NON comunicante

Cisti interemisferica

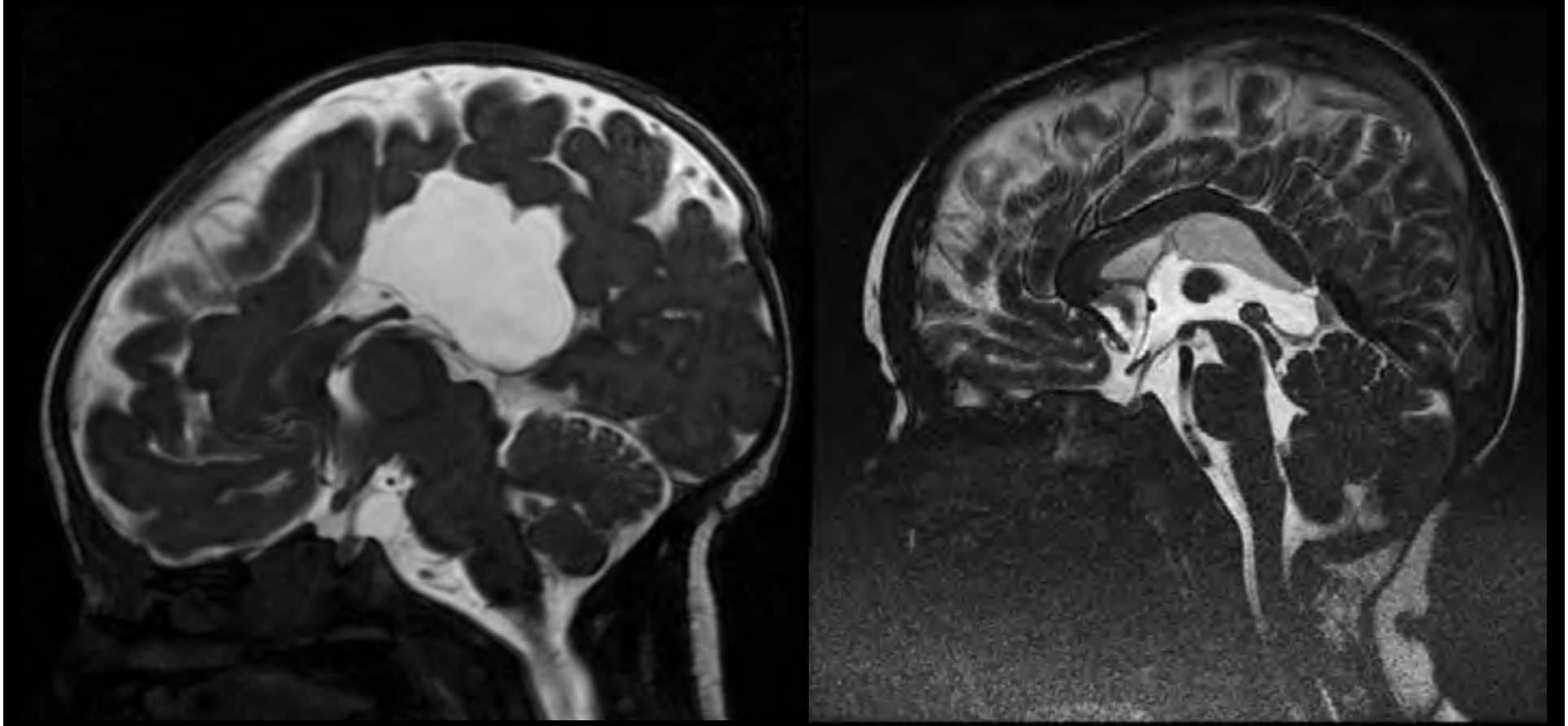
Acta Paediatr 2005;94:1066-1072

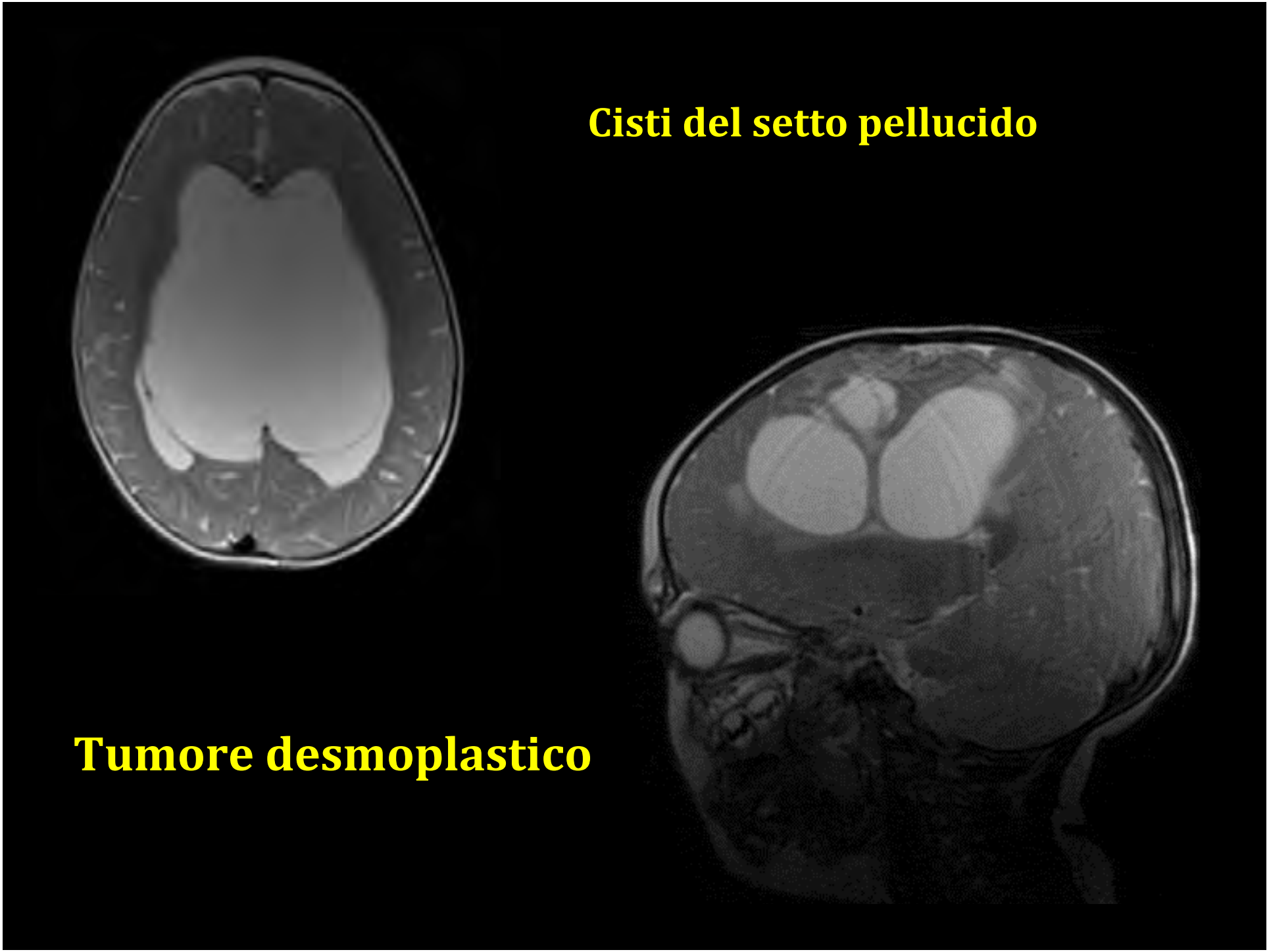
CISTI INTEREMISFERICHE
Comunicanti con i ventricoli → tipo 1





CISTI INTEREMISFERICHE
NON comunicanti con i ventricoli → tipo 2




Cisti del setto pellucido
Tumore desmoplastico

MALFORMAZIONI delle COMMISSURE CEREBRALI

AGENESIA delle COMMISSURE con LIPOMI INTEREMISFERICI

Cisterna cerebellopontina

DISTURBO DIFFERENZIAZIONE
delle MENINGI primordiali

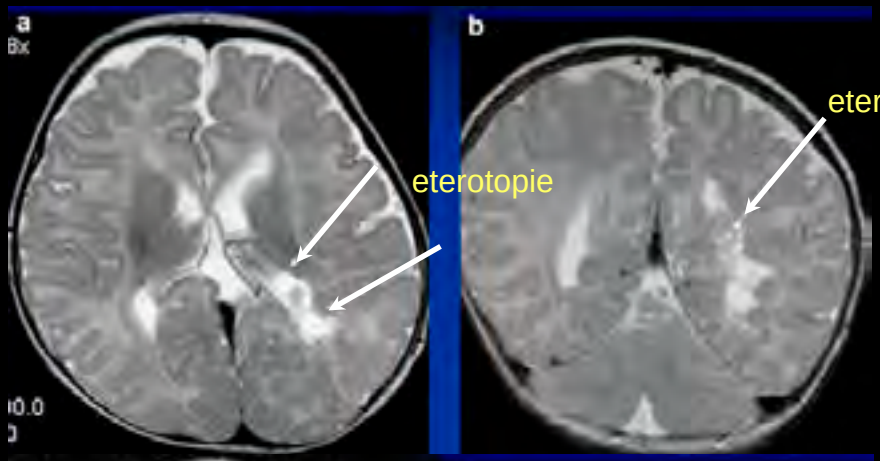
Mesenchima MENINGI -->
Differenzia

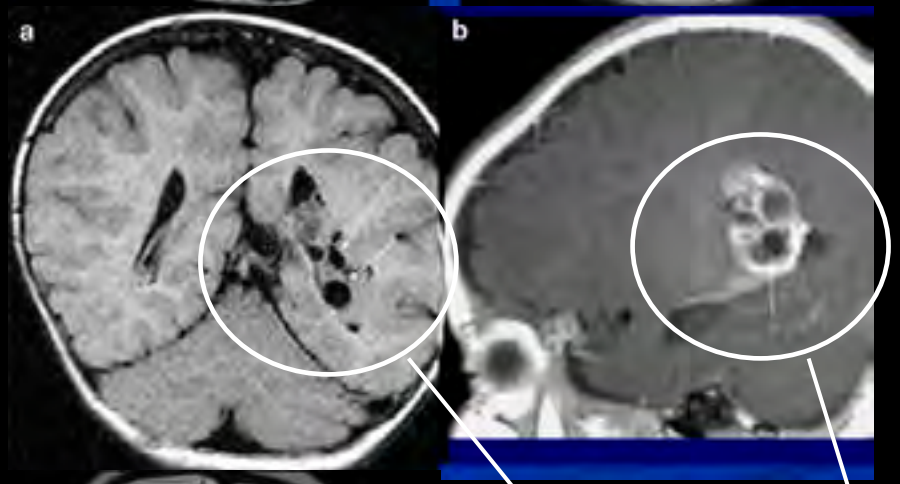
SEDE subaracnoidea

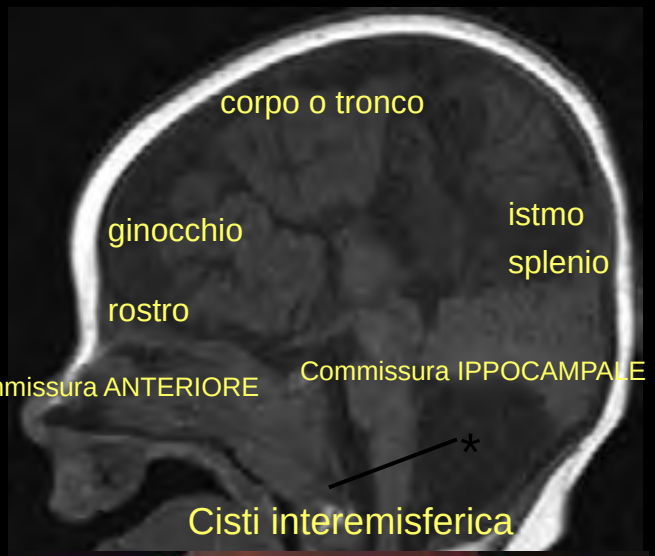
- fissura interemisferica (40%)
- cisterna sopracerebellare (25%)
- cisterna interpeduconlare (20%)
- cisterna cerebellopontina (10%)
- cisterna silviana (5%)

Lipomi della linea mediana → falce, ipotalamo, tectum mesencefalico

Sindrome di AICARDI

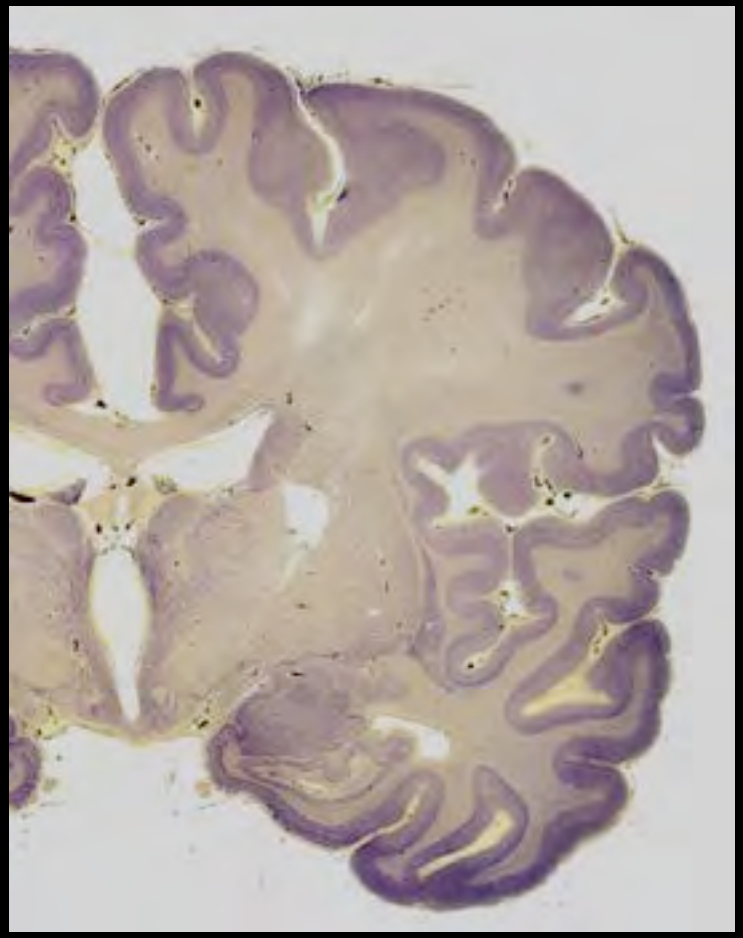


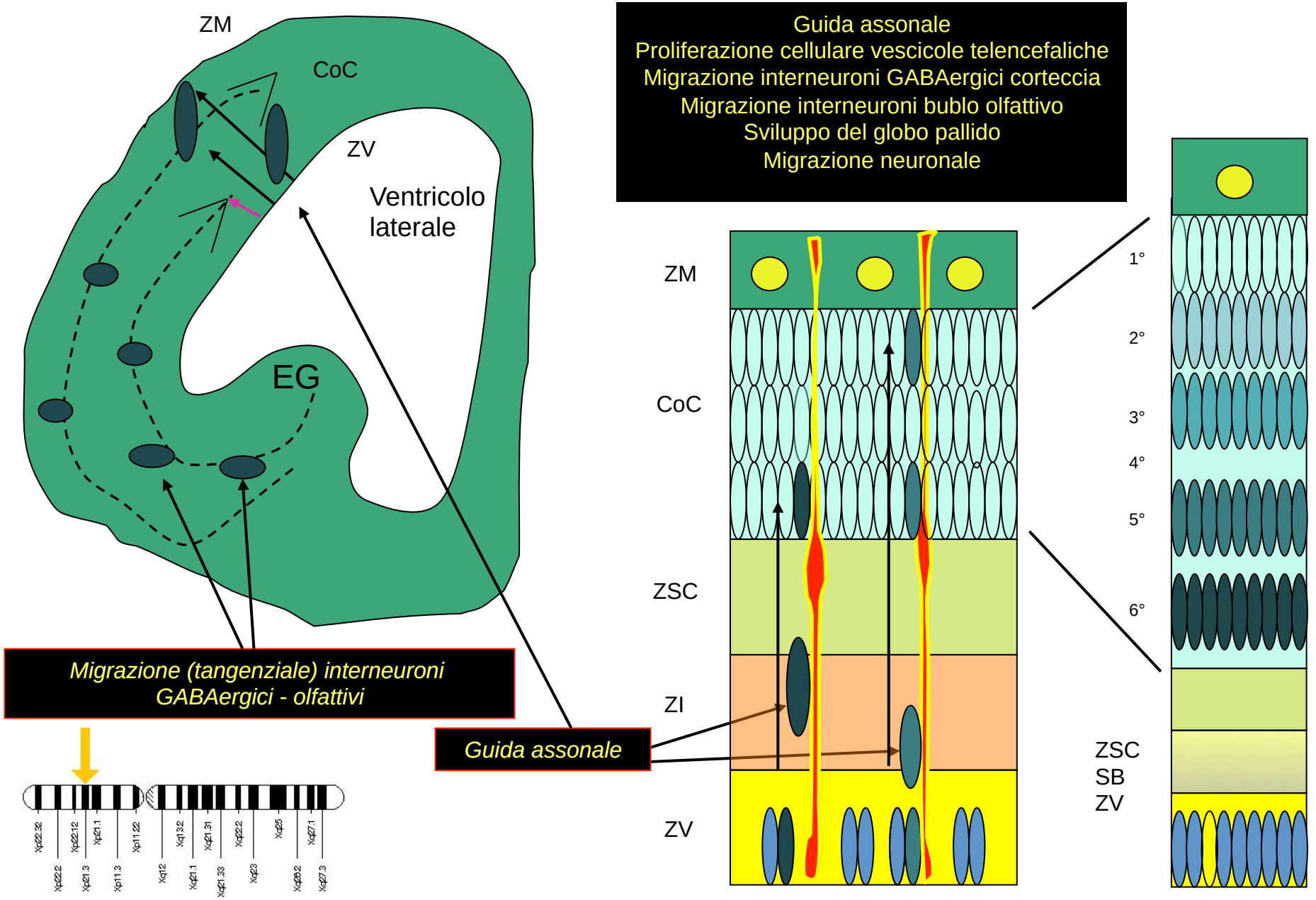


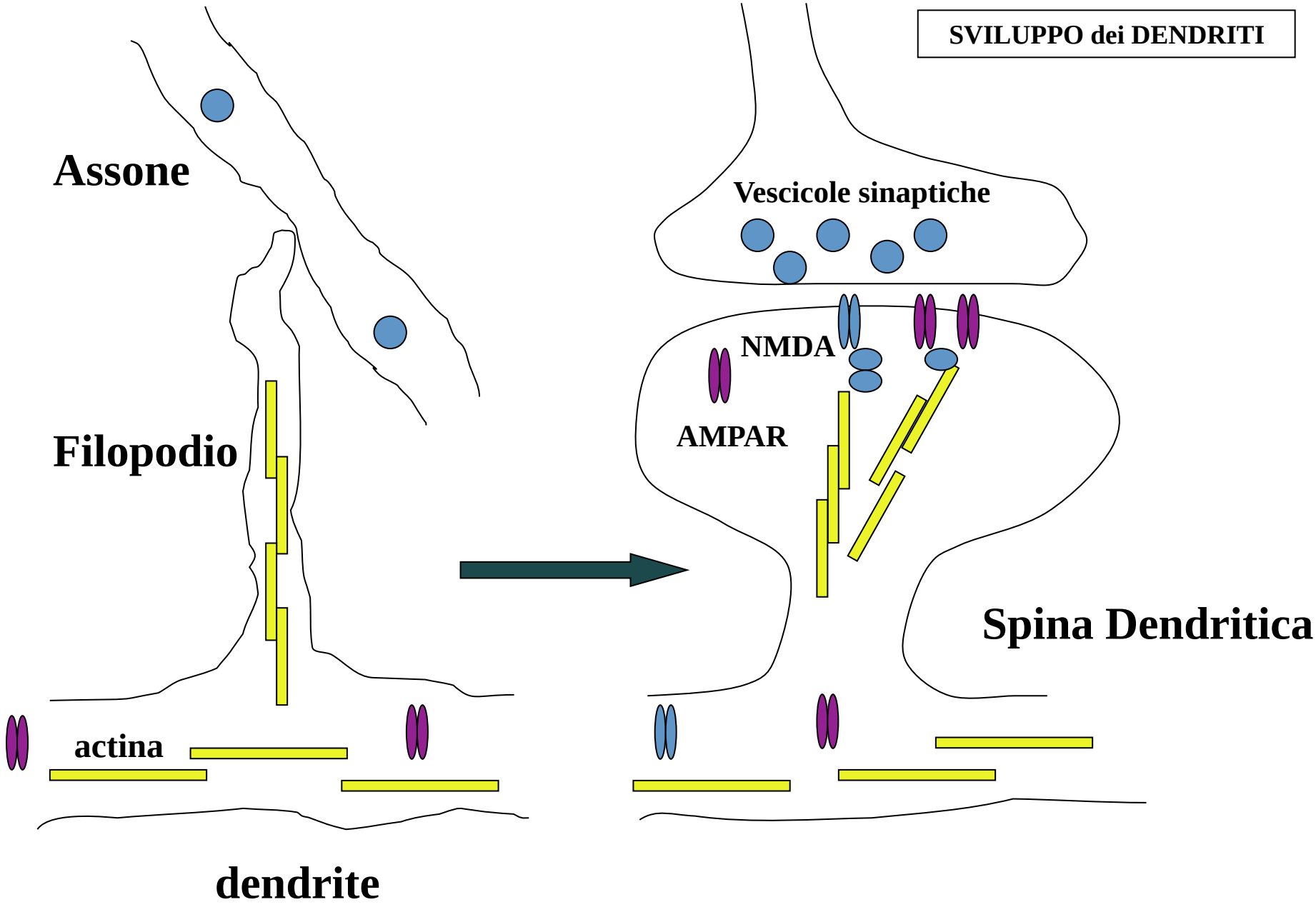


Brain & Dev 2004;67:107-109

QUALCOSA VIENE “COSTRUITA”MALE ….



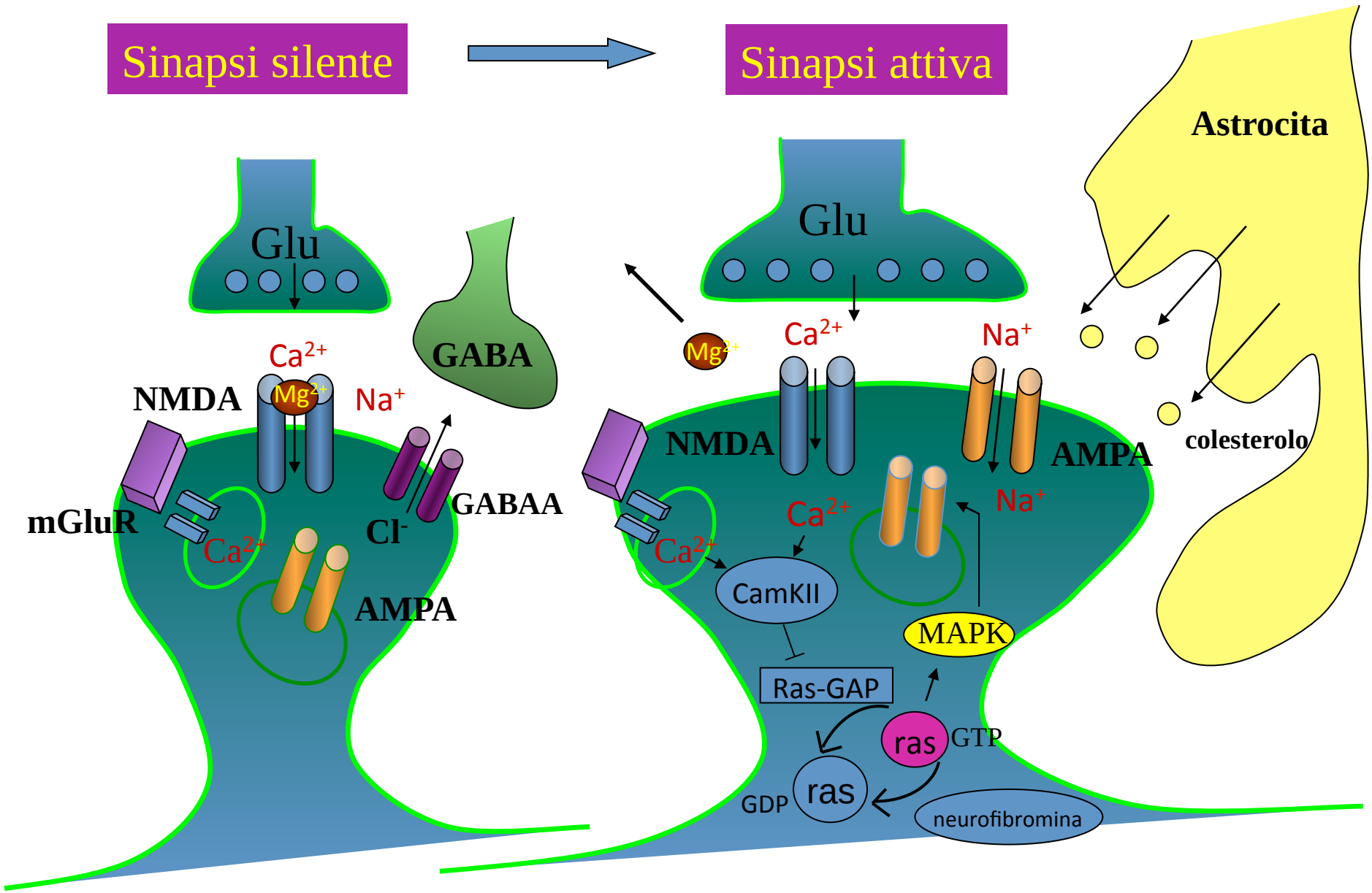

ZM
CoC
ZV
Ventricolo laterale
EG
Migrazione (tangenziale) interneuroni GABAergici - olfattivi
Guida assonale
Guida assonale
Proliferazione cellulare vescicole telencefaliche
Migrazione interneuroni GABAergici corteccia
Migrazione interneuroni bublo olfattivo
Sviluppo del globo pallido
Migrazione neuronale
ZM
CoC
ZSC
ZI
ZV
1°
2°
3°
4°
5°
6°
ZSC
SB
ZV



Modificato da Catania MV. Neurobiologia dello Sviluppo. In: Pavone L, Ruggieri M. *Neurologia Pediatrica*. 2° ed. Elsevier/Masson, 2006: p. 77

Modificato da Catania MV. Neurobiologia dello Sviluppo. In: Pavone L, Ruggieri M. *Neurologia Pediatrica*. 2° ed. Elsevier/Masson, 2006: p. 80

FORMAZIONE e MATURAZIONE delle SINAPSI



MALFORMAZIONI dello SVILUPPO CORTICALE

I. MALFORMAZIONI DA ANOMALIE DELLA PROLIFERAZIONE/APOPTOSI DEI PRECURSORI NEURONALI E GLIALI

A) DIMINUITA PROLIFERAZIONE - AUMENTATATA PROLIFERAZIONE/DIMINUITA APOPTOSI DEI PRECURSORI NEURONALI E GLIALI

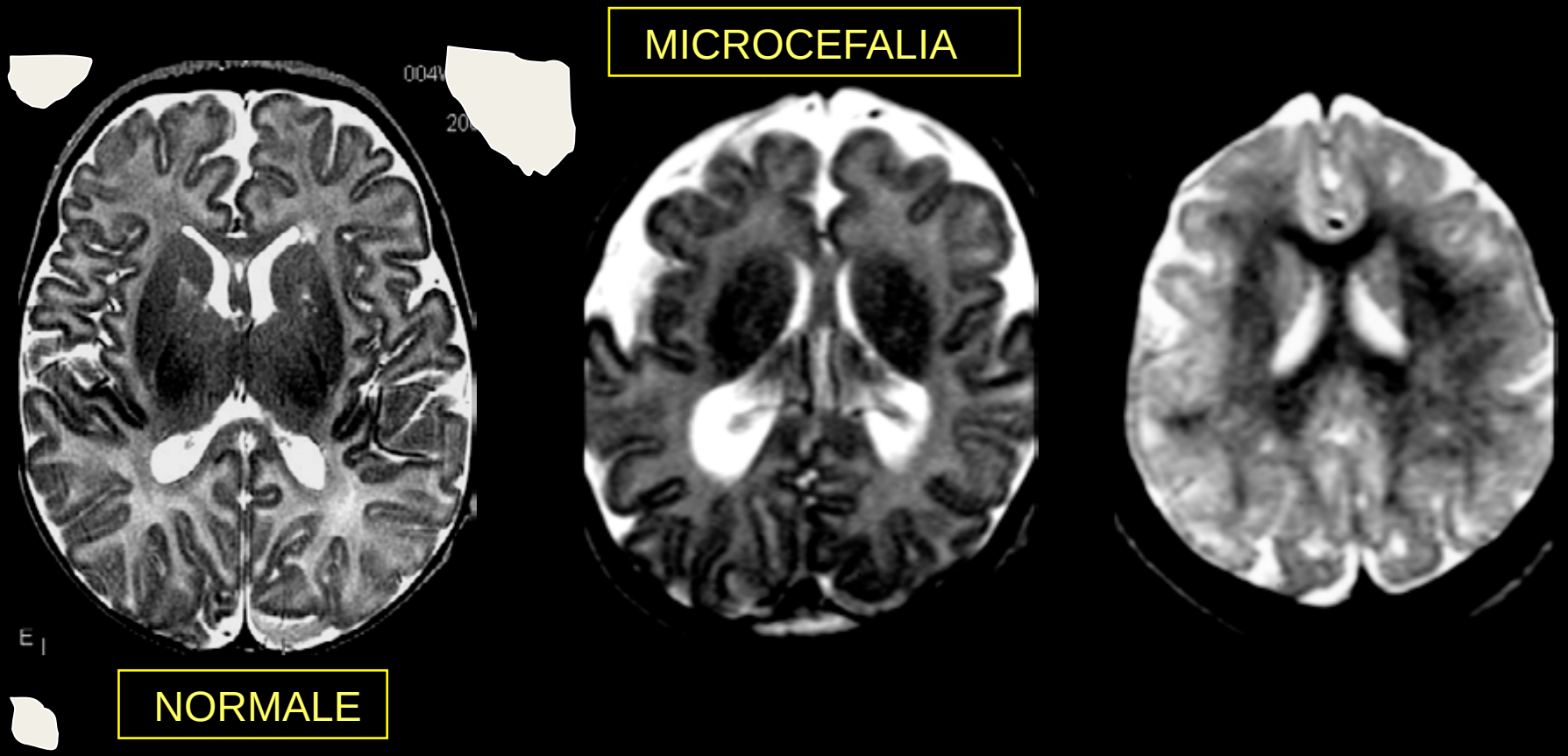
Anomalie delle dimensioni cerebrali





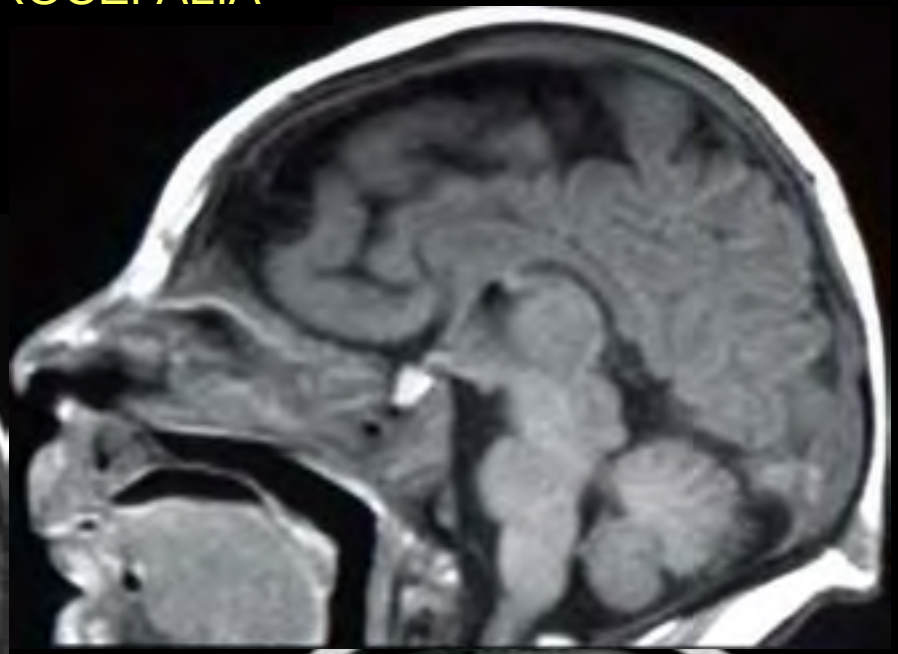


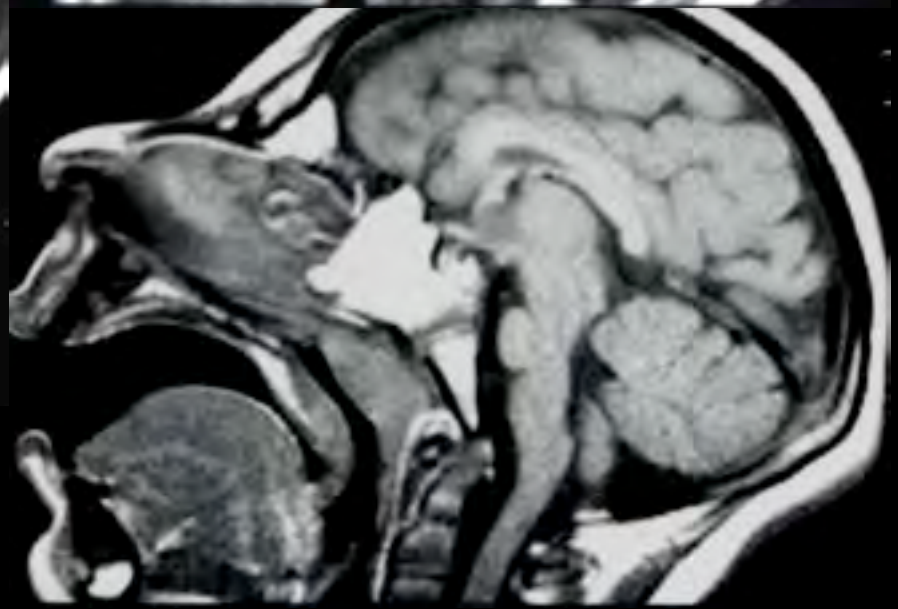
MICROCEFALIA
OLIGOGIRIA



MICROCEFALIA

NORMALE





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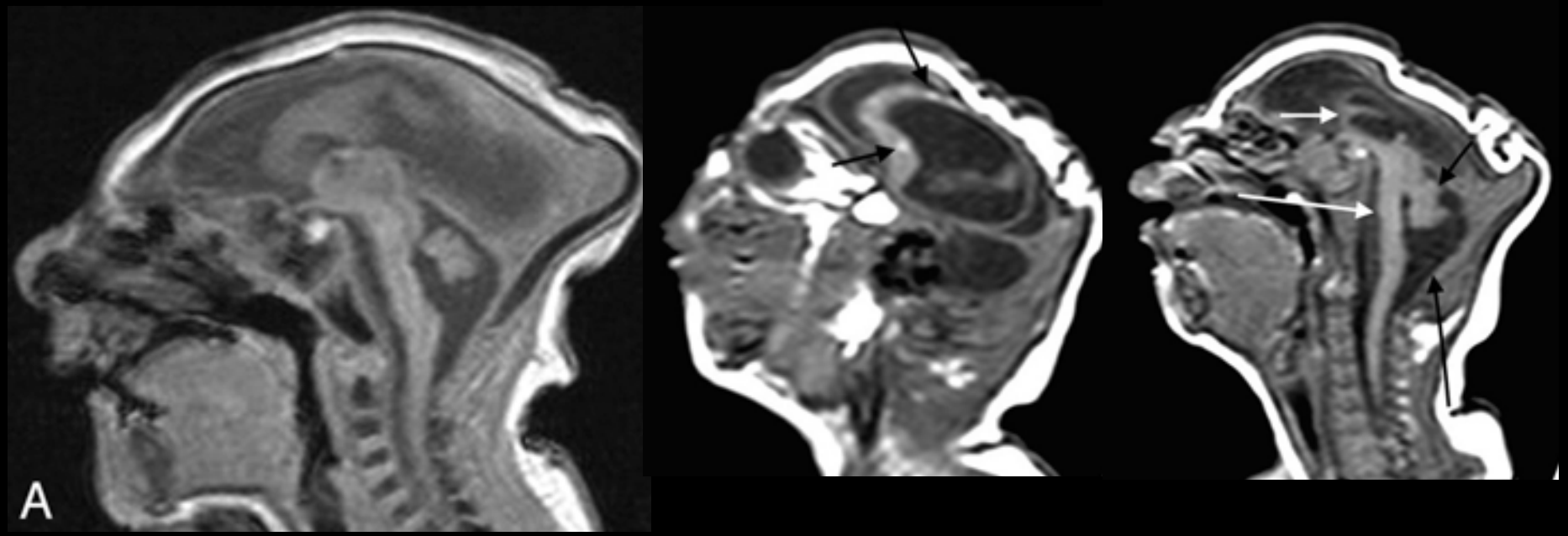
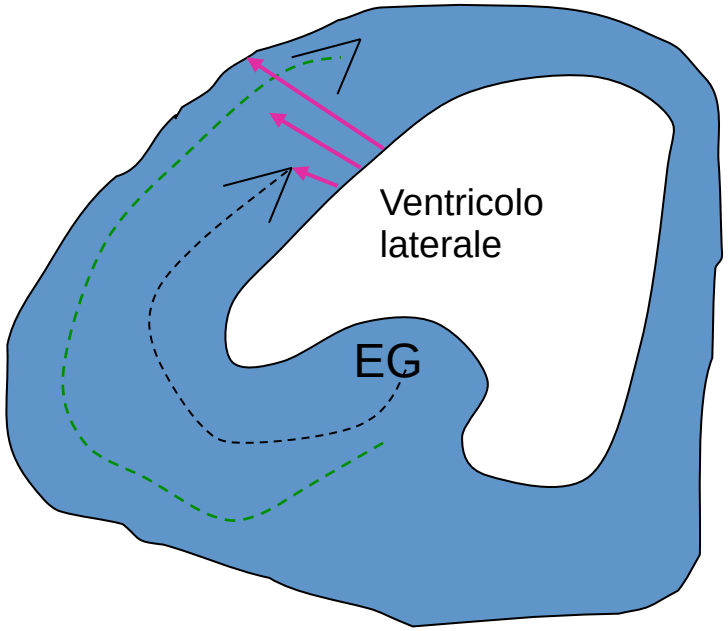
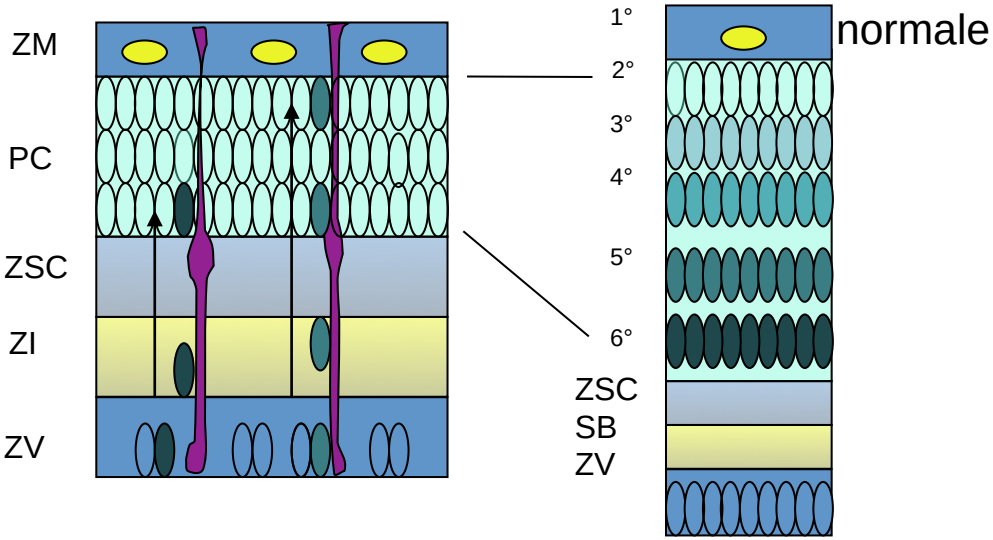
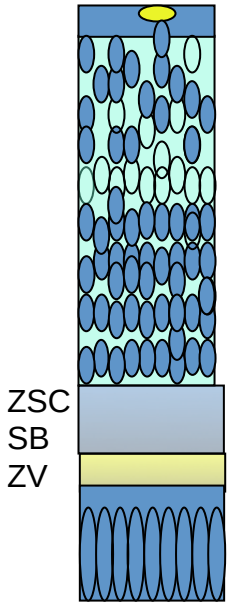






Lissencefalia



Modificato da Catania MV. Neurobiologia dello Sviluppo. In: Pavone L, Ruggieri M. *Neurologia Pediatrica*. 2° ed. Elsevier/Masson, 2006: p. 73

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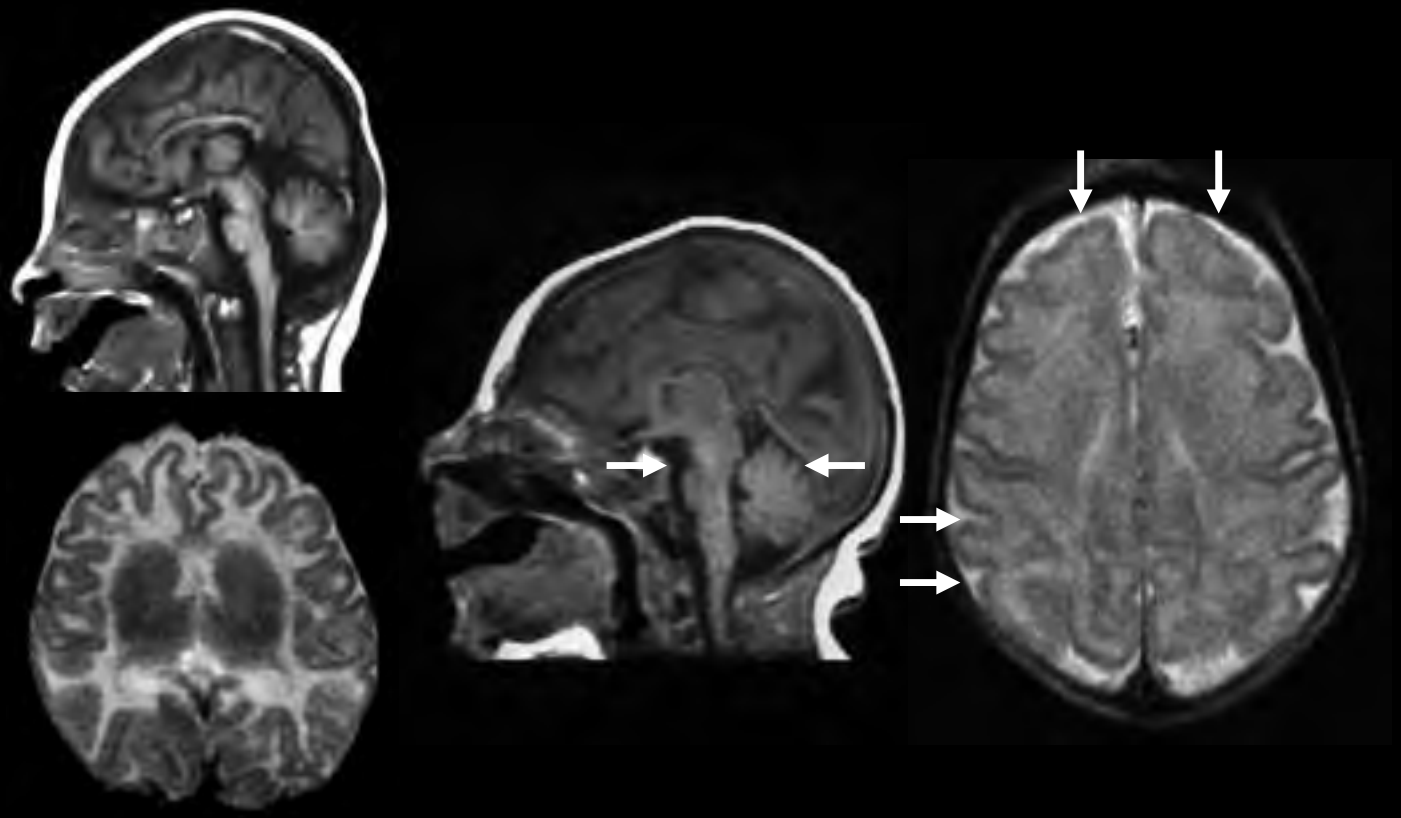
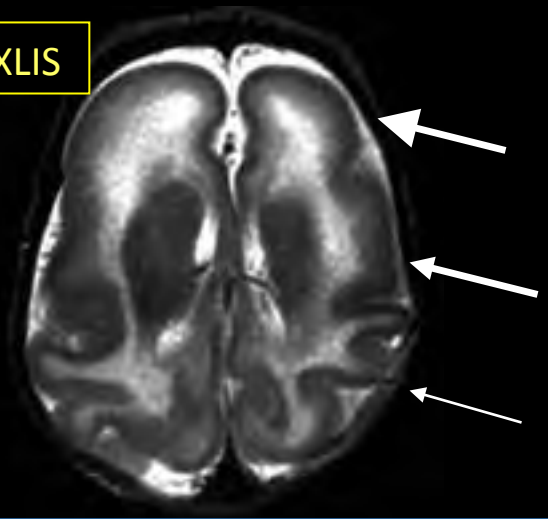


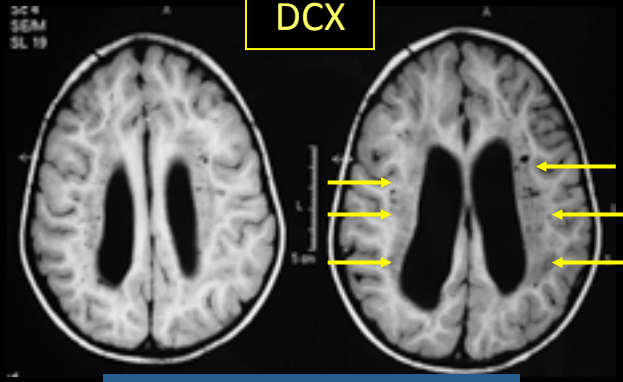
Lissencephaly (LISX1, XLIS), Doubke cortex (DCX)
X-linked double cortex syndrome
Subcortical laminar heterotopia (SCLH)
X-linked subcortical band heterotopia (SBX)
[LISX1, XLIS, SCLH, SBH; MIM # 300067]

Lissencephaly (LIS1)
Classic lissencephaly sequence
Isolated subcortical laminar heterotopia (SCLH)
Subcortical band heterotopia (SBH)
[LIS1, SCLH, SBH; MIM # 300067]

XLIS

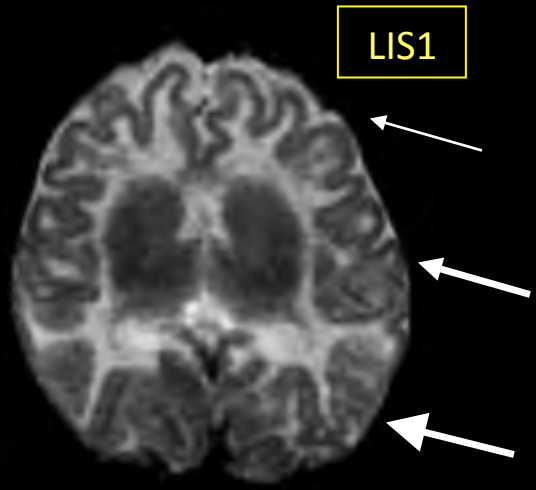


DCX



Doppia corteccia (DCX)

LIS1



Lissencefalia (LISX1 - XLIS)
[gradiente frontale >> parietale > occipitale

Lissencefalia (LIS1)
[gradiente parieto-occipitale >> frontale

MALFORMAZIONI dello SVILUPPO CORTICALE

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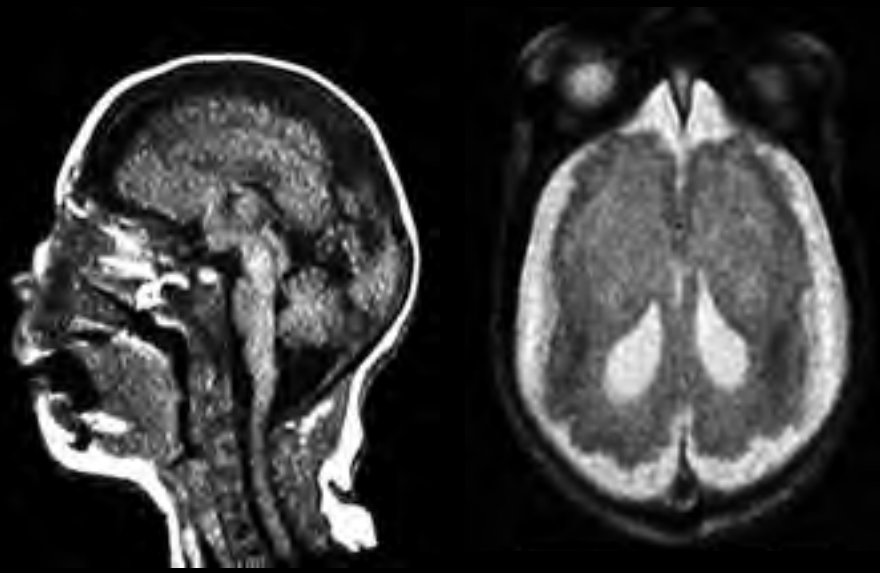
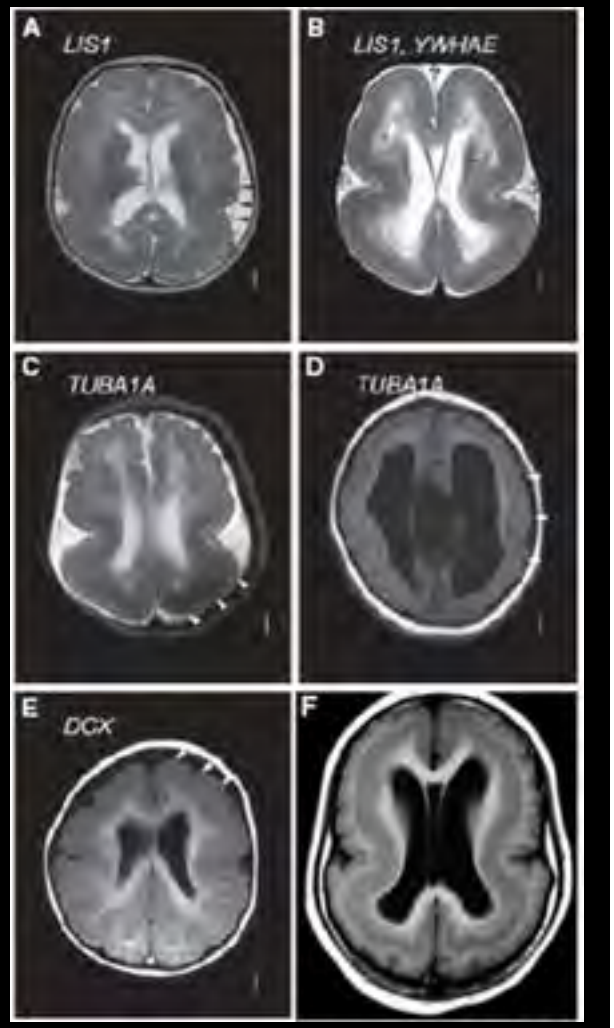




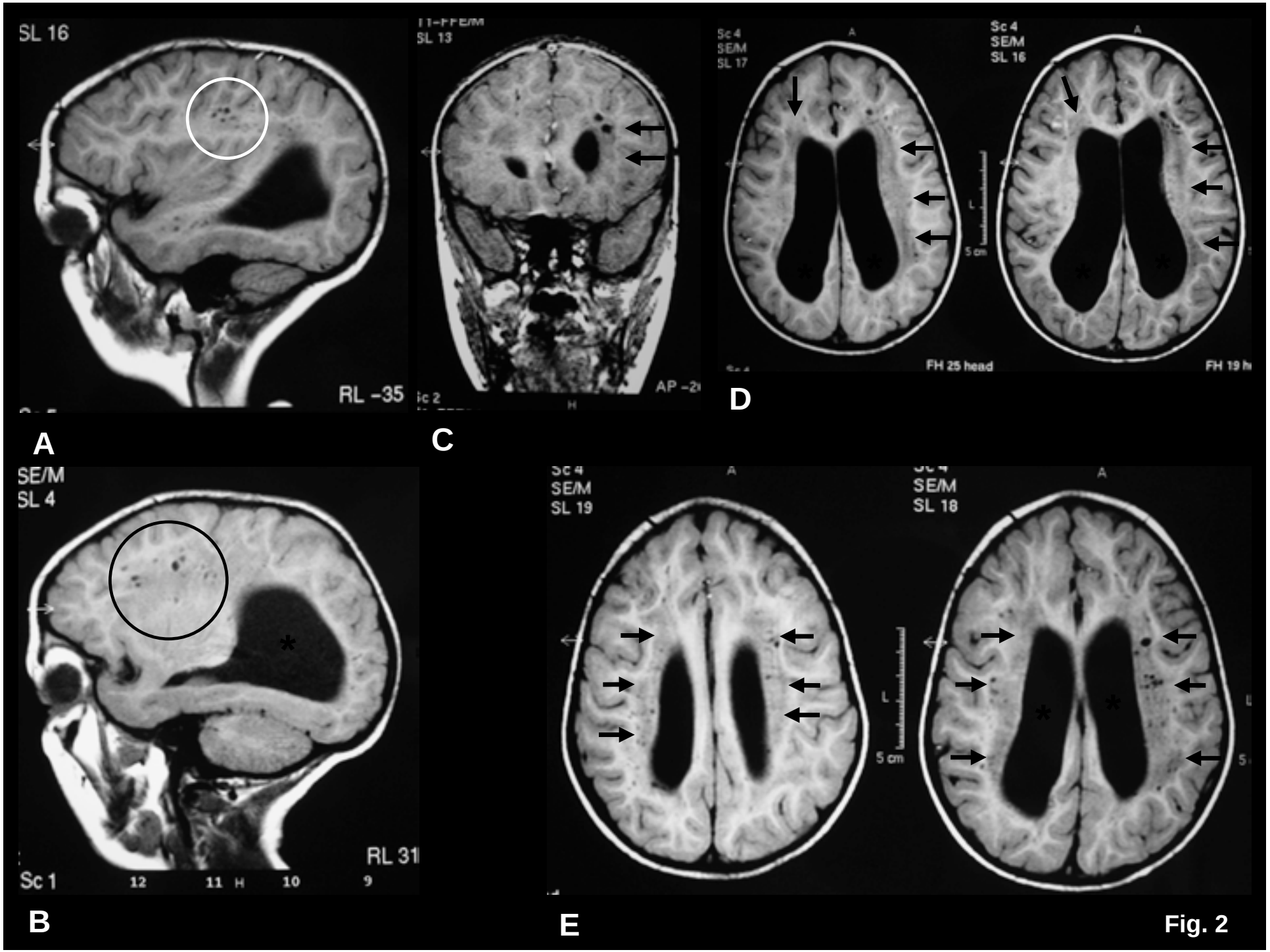
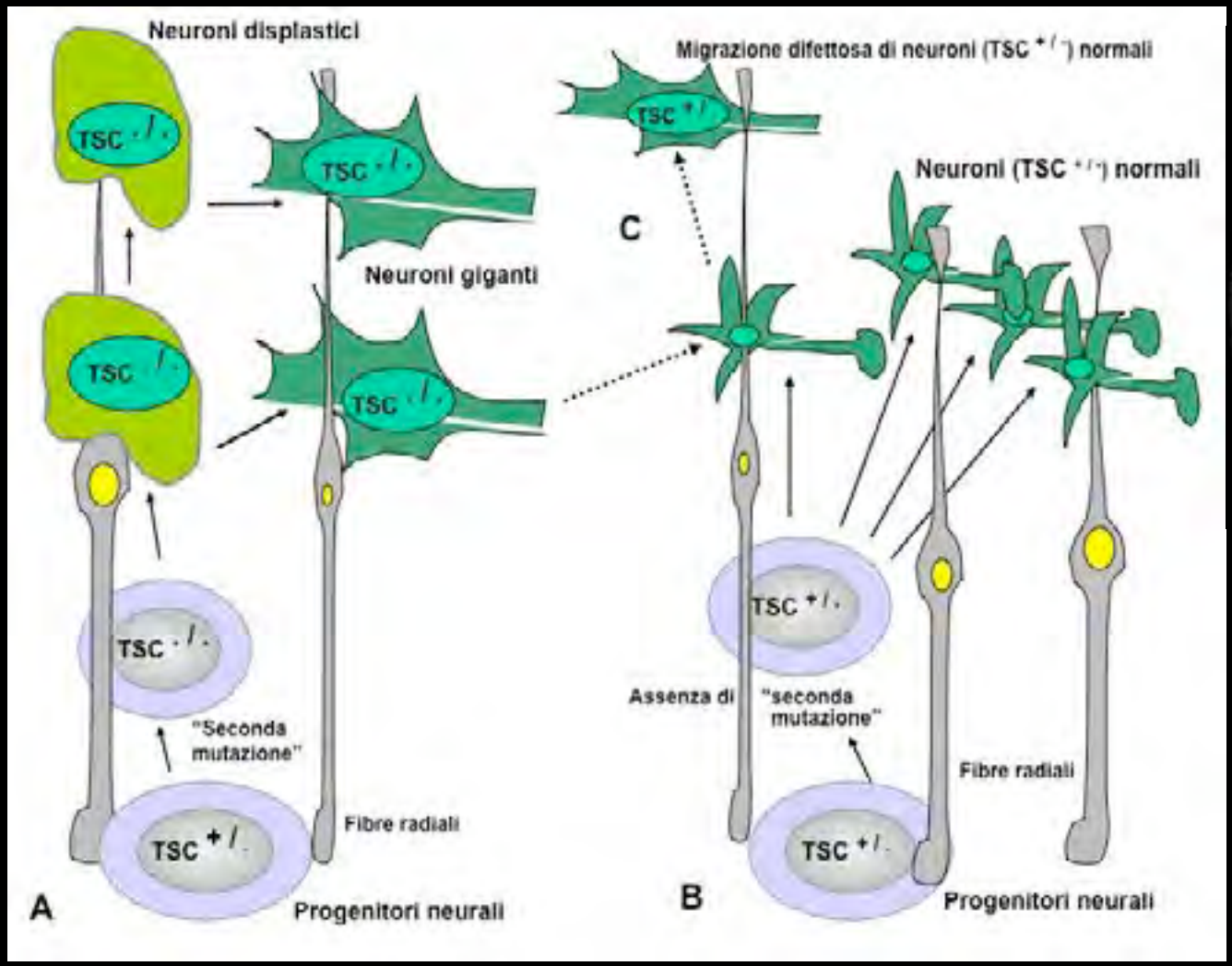

SL 16
RL -35
A
SL 13
AP -2
C
Sc 4
SE/M
SL 17
Sc 4
SE/M
SL 16
FH 25 head
FH 19 h
D
SE/M
SL 4
RL 31
Sc 1
12
11
10
9
B
SE/M
SL 19
SE/M
SL 18
5 cm
E

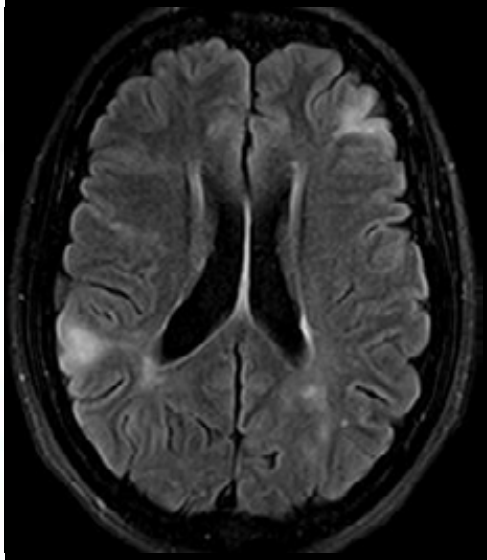
Fig. 2

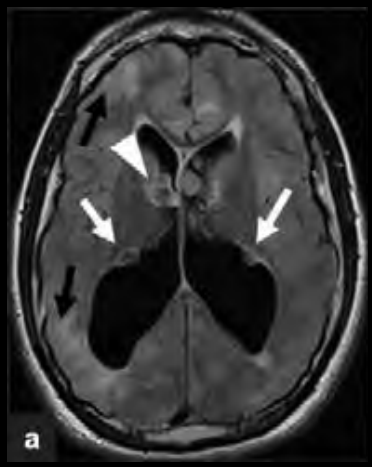
MALFORMAZIONI dello SVILUPPO CORTICALE

I. MALFORMAZIONI DA ANOMALIE DELLA PROLIFERAZIONE/APOPTOSI DEI PRECURSORI NEURONALI E GLIALI

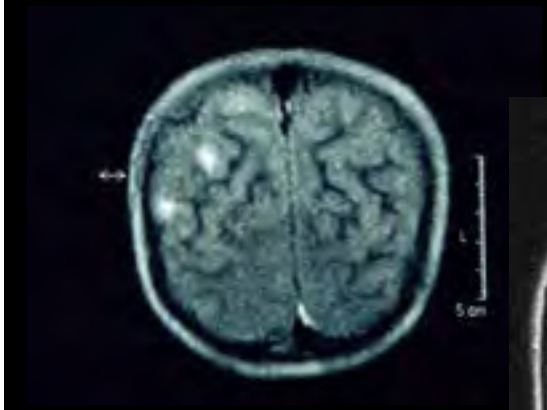
A) PROLIFERAZIONE ANOMALA (TIPI CELLULARI ANOMALI)

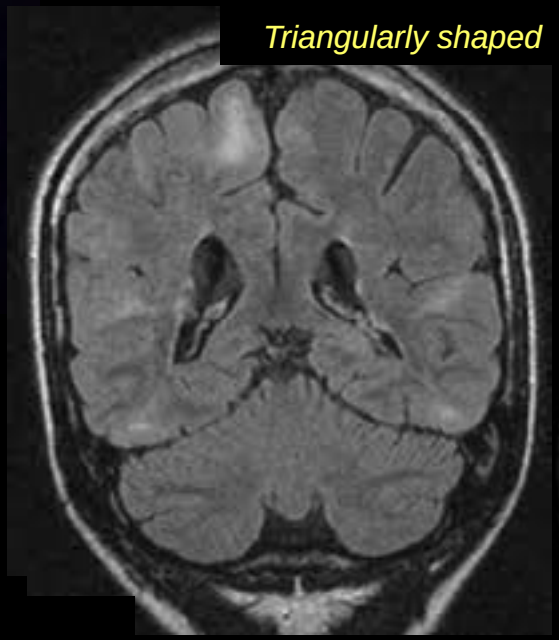




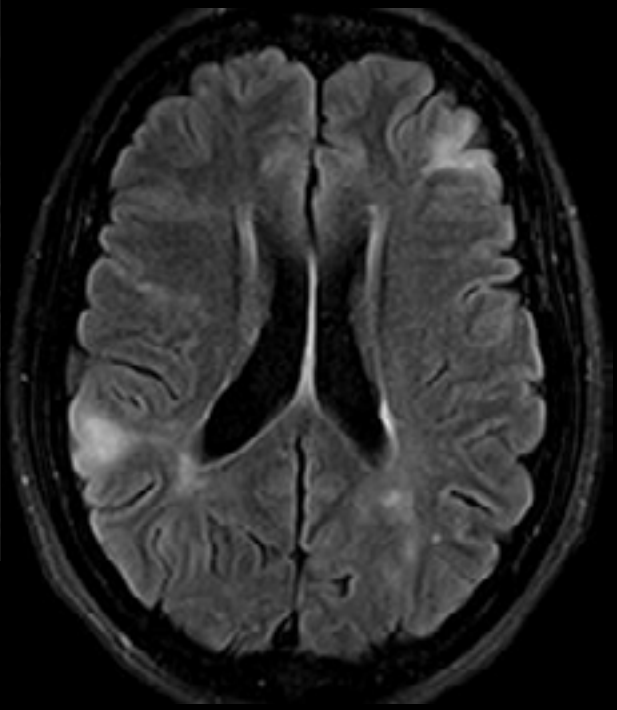


Cortical tubers (98 %)





Triangularly shaped



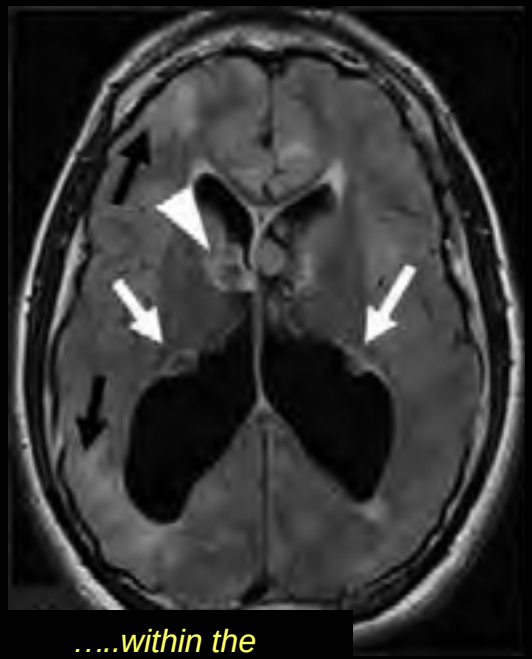
Triangularly shaped

Cortical - subcortical

subcortical

…… follow the contour of circonvolutions….

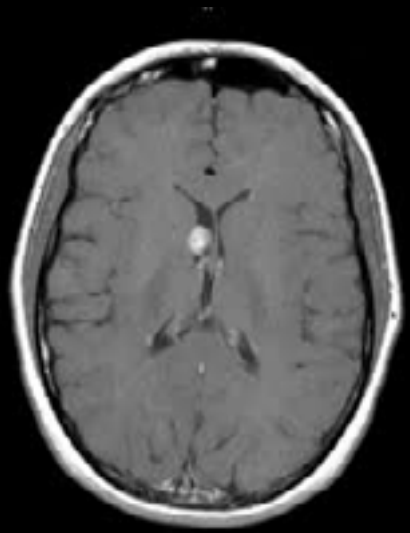
Subependymal nodules(95 %)

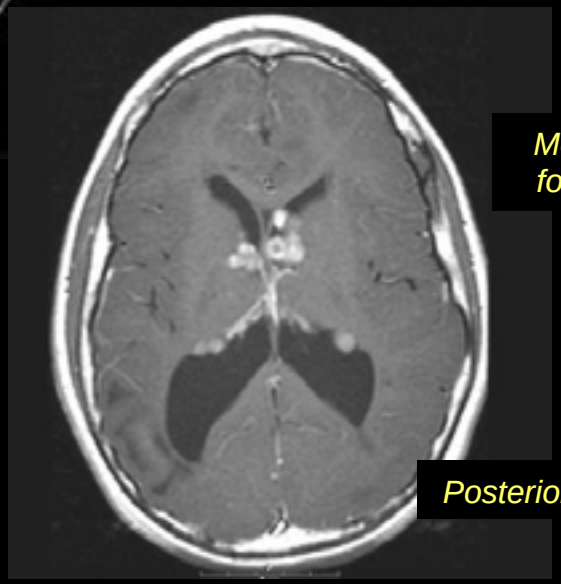


…..within the ependymal layer…

…… calcified [TC]….

Monroe's foramina

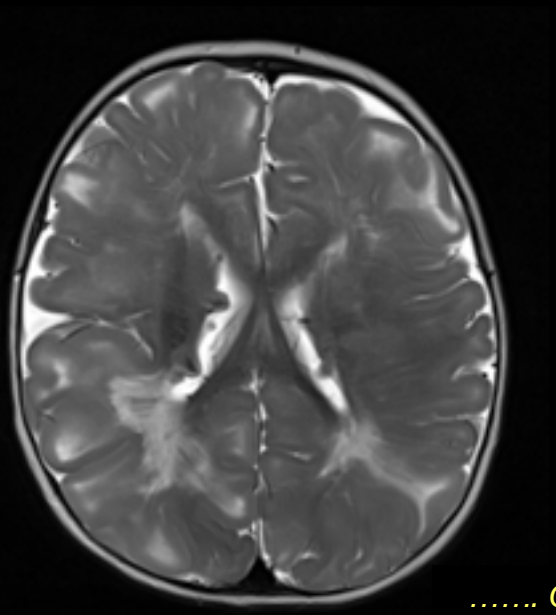




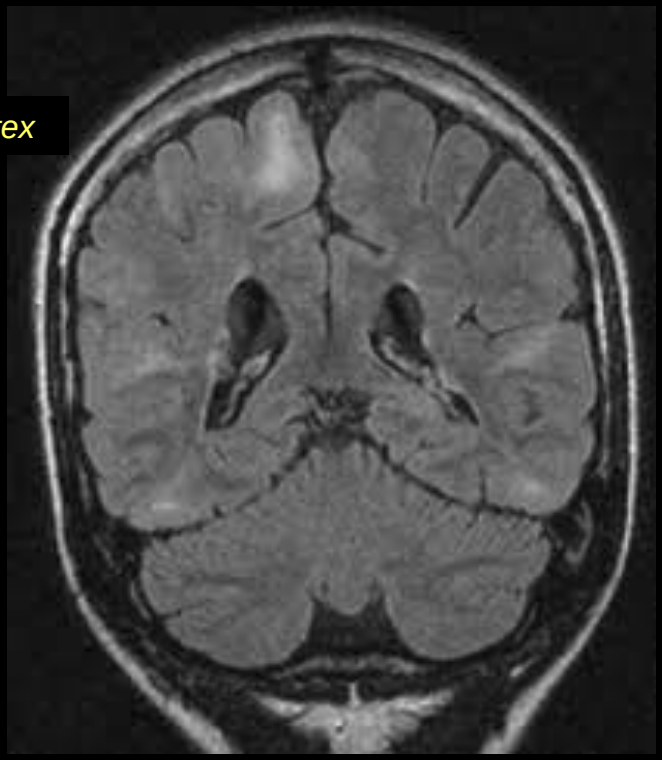
Monroe's foramina

Posterior ventricular cells

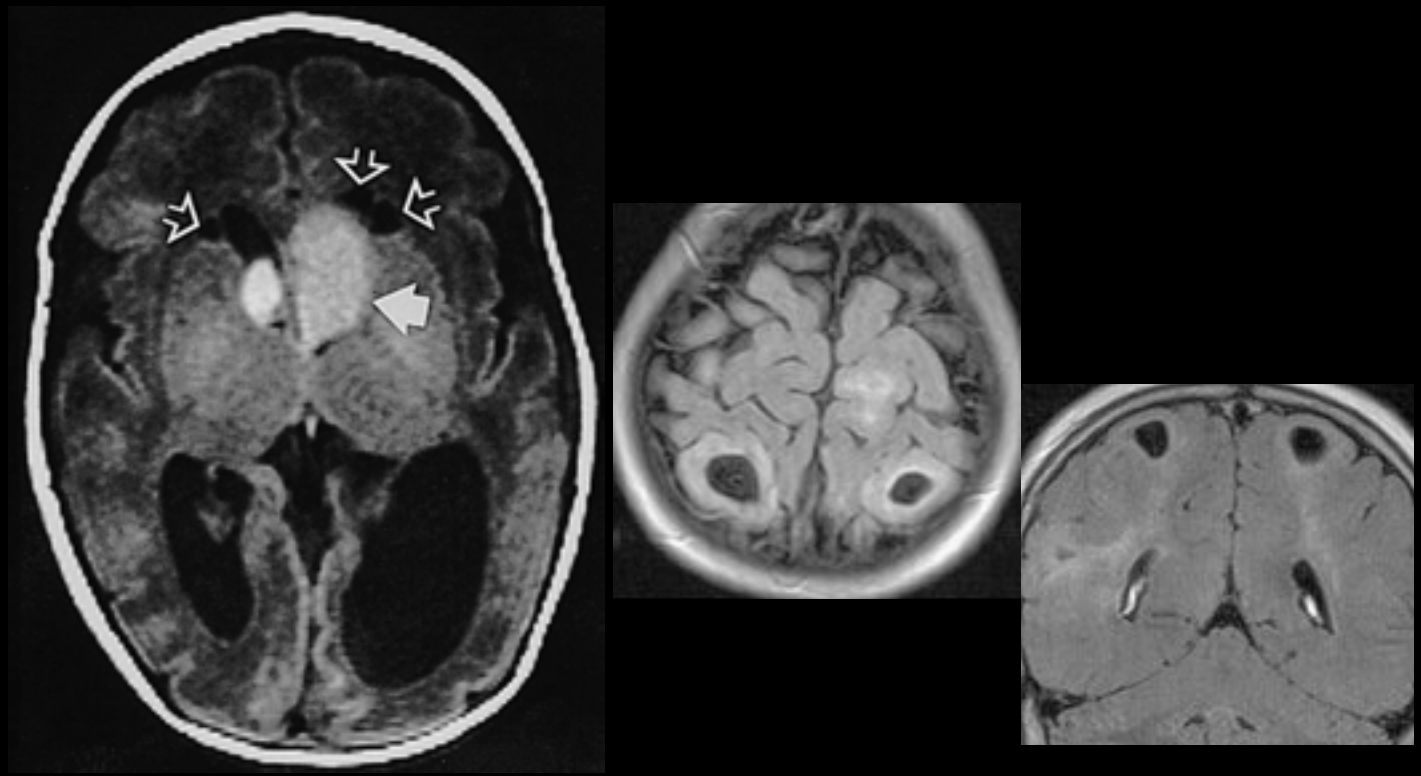
White matter linear streaks
(93 %)

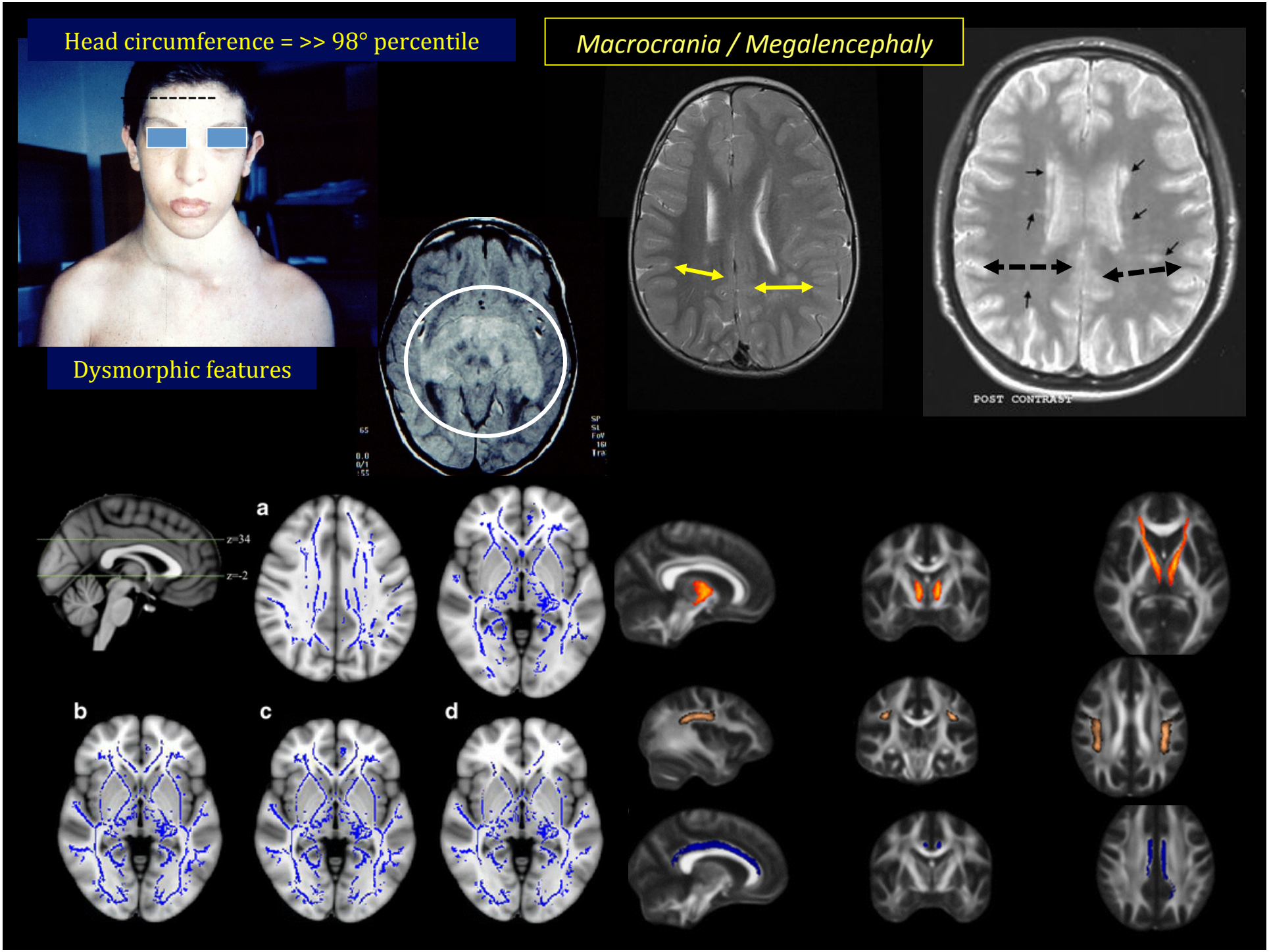


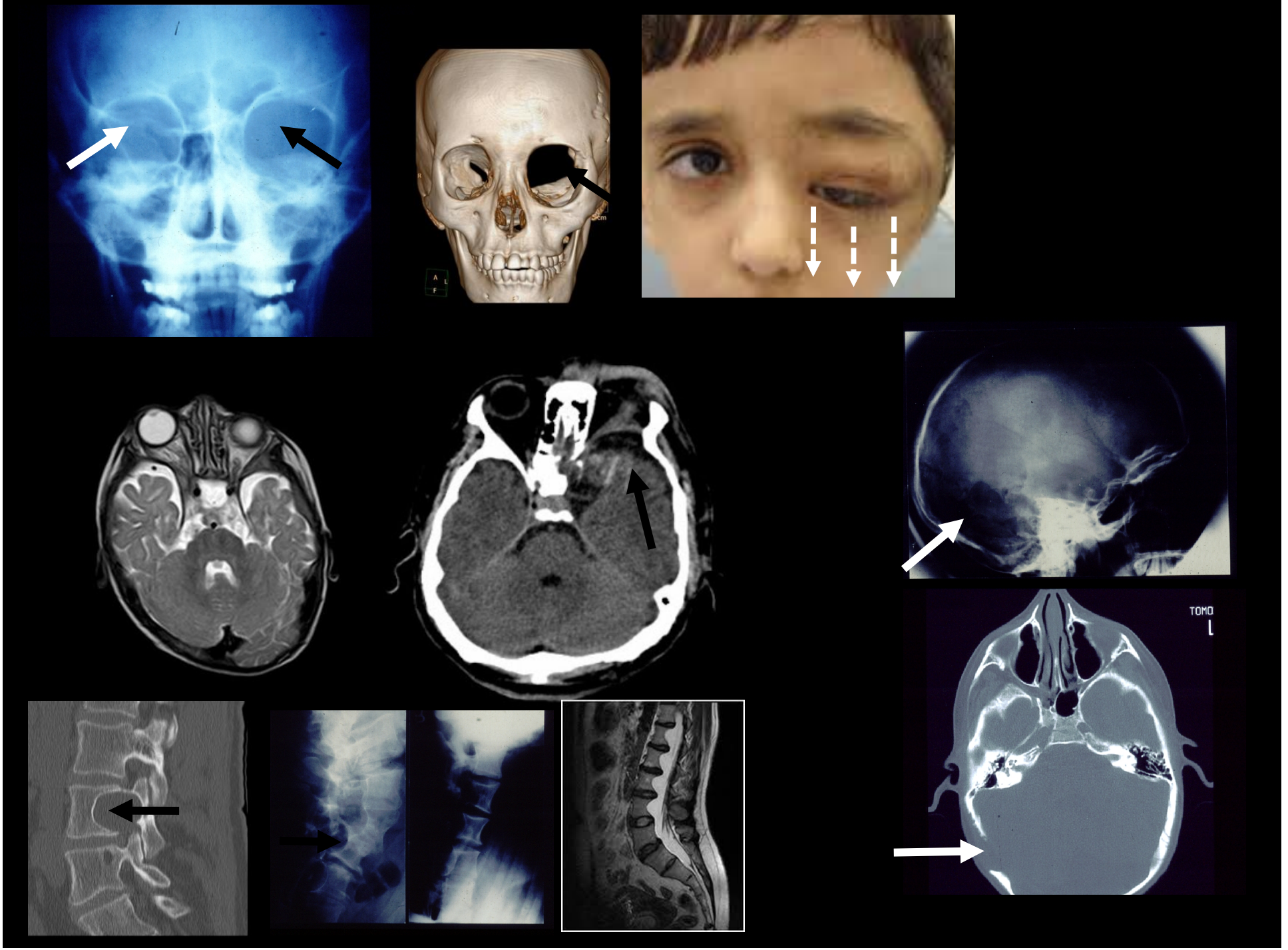
……. Oriented from ventricles → towards cortex



Cerebral cysts (5 - 10 %)




Head circumference = >> 98° percentile
Dysmorphic features
Macrocrania / Megalencephaly
POST CONTRAST
z=34
z=-2
a
b
c
d


TOMO
L

Legius syndrome [NF1-like syndrome]

Café-au-lait spots (atypical)

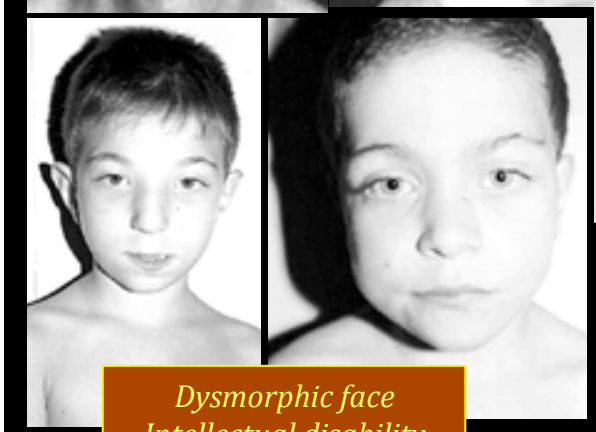
Macrocrania / Megalencephely

lipomas

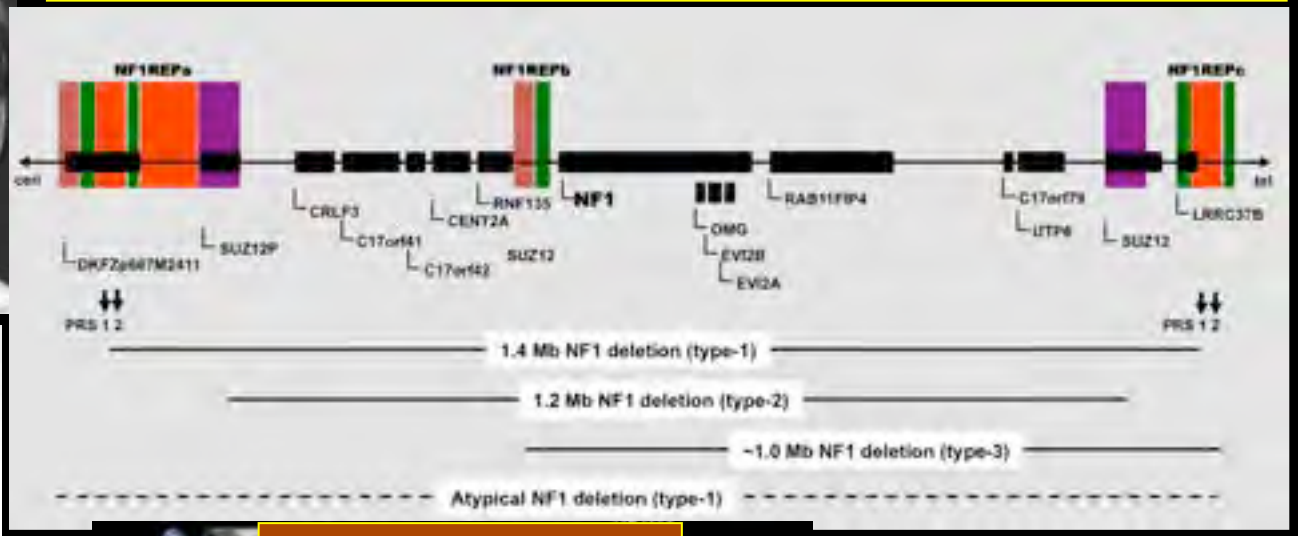
BEHAVIOURAL/NEUROLOGICAL PHENOTYPE

Learning disabilities
Attention deficit / hyperactivity
Cognitive delay [IQ <<]
Behavioural disturbances (oppositive)
Epilepsy

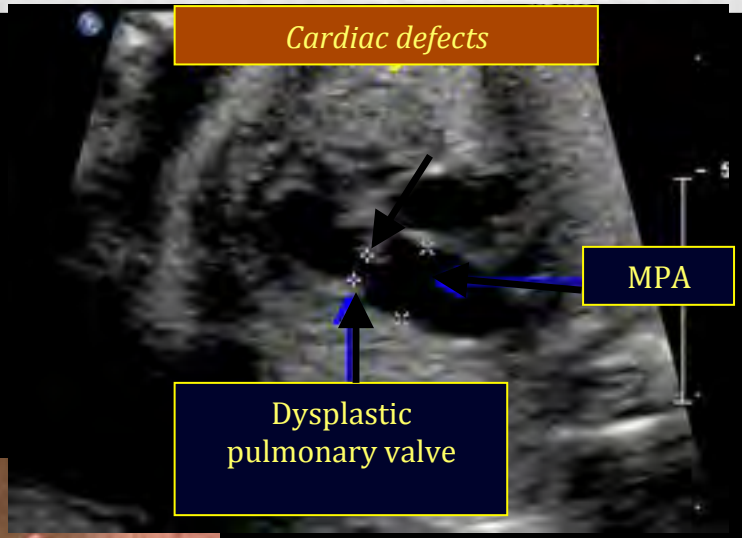
NF1 gene - "*microdeletion syndrome*" [4.2 %]



Dysmorphic face
Intellectual disability

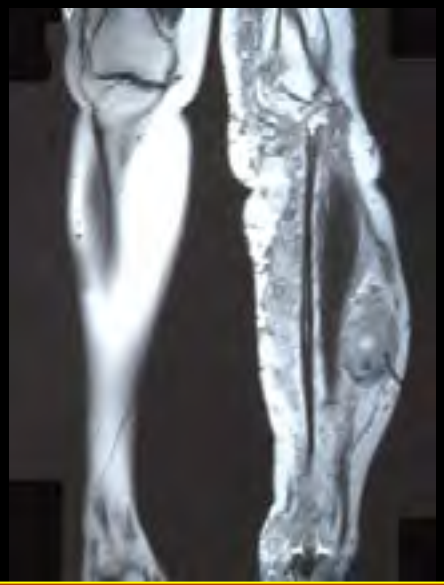


Cardiac defects

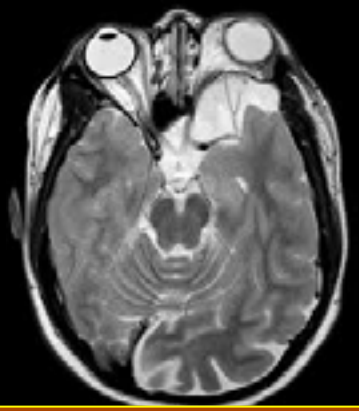


MPA

Dysplastic
pulmonary valve



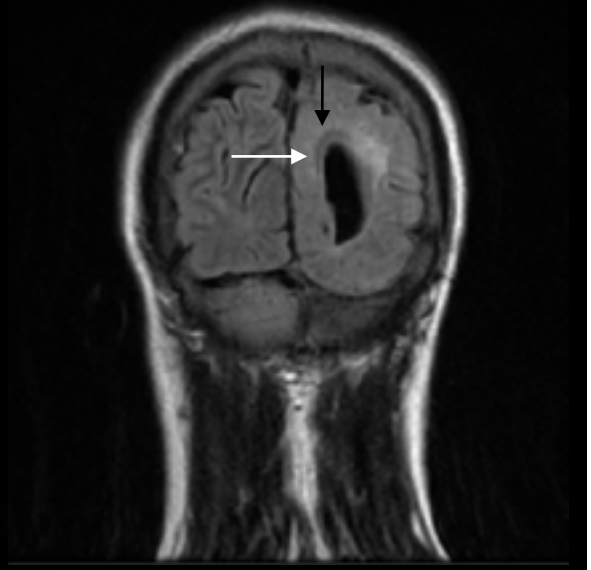
>> MPNST (over time)

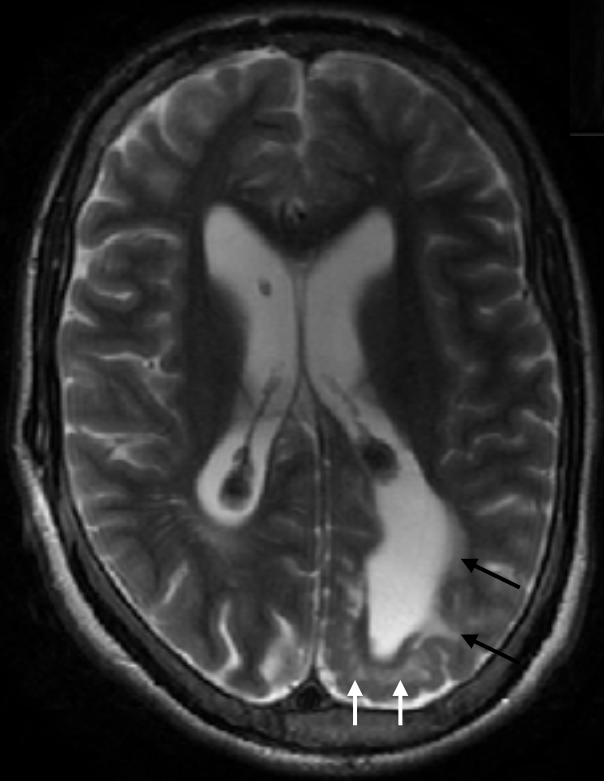


Sphenoid bone dysplasia

Neurofibromas → younger ages

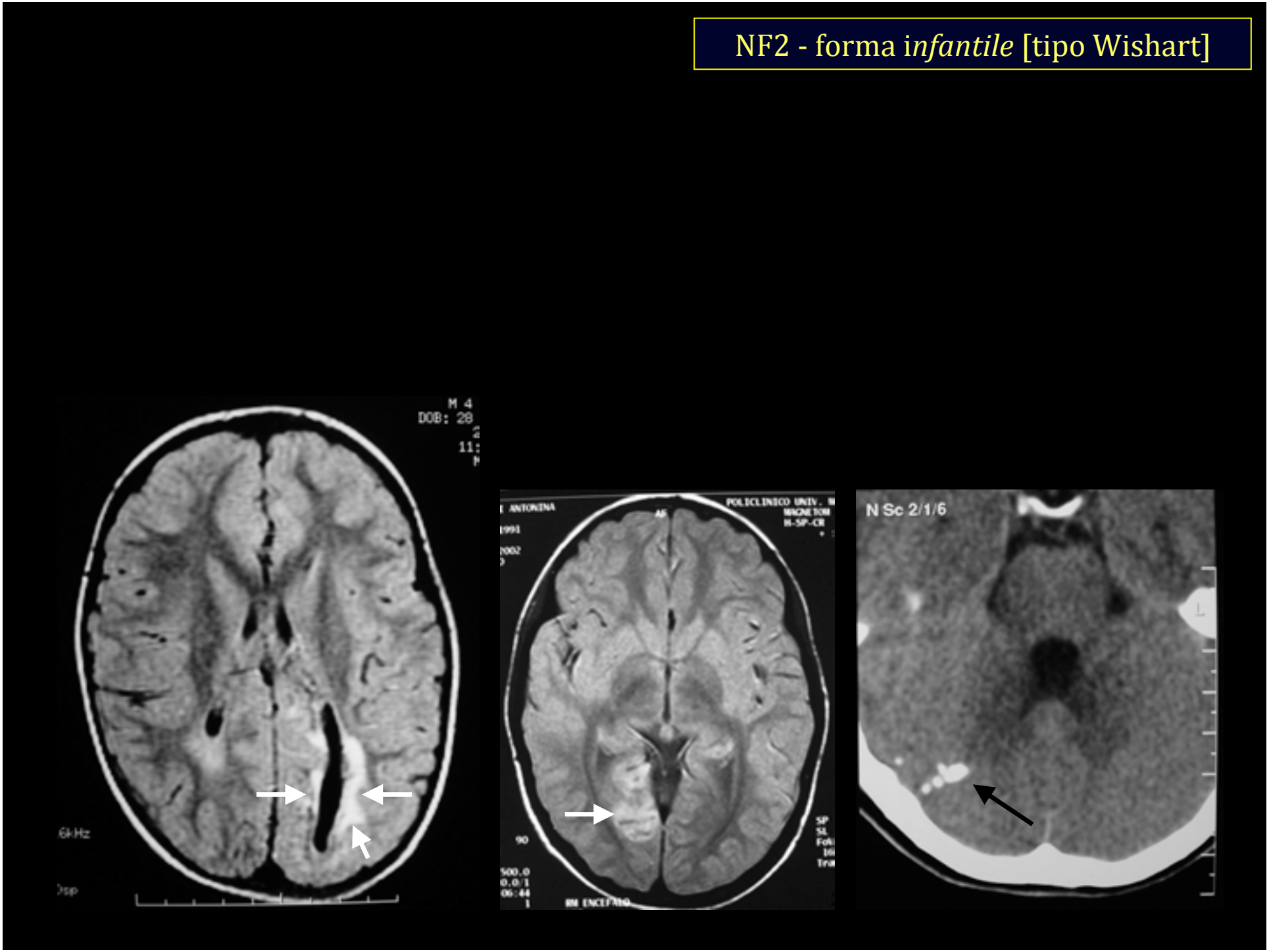
NF2 - forma "*congenita*"



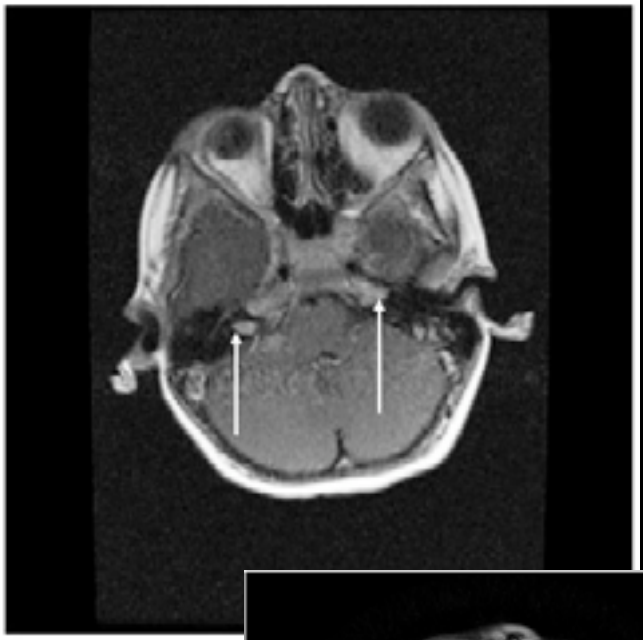


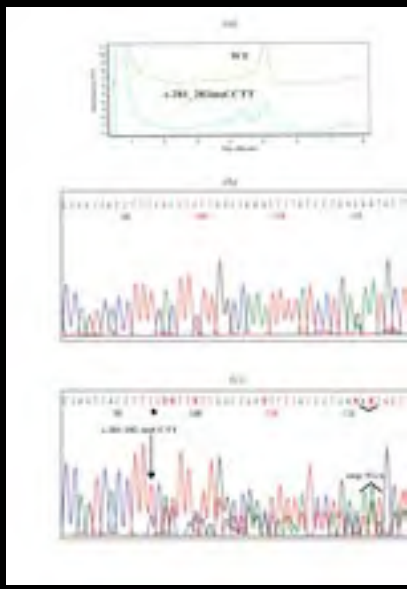
Malformazioni dello sviluppo corticale

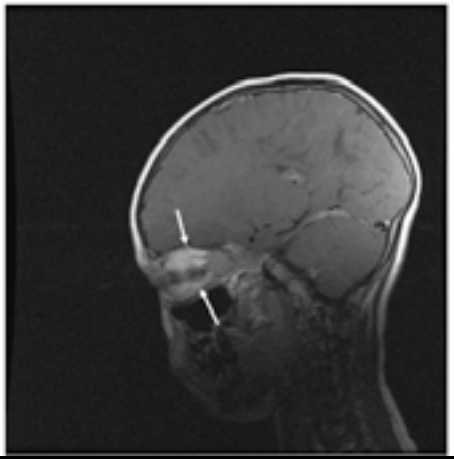
NF2 - forma i*nfantile* [tipo Wishart]



NF2 - forma "*congenita*"



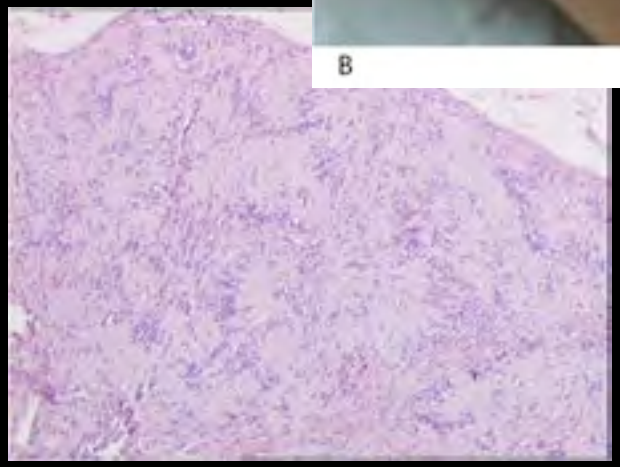




NF2 - "*Congenital" form*

A

B



C

Sturge-Weber syndrome

4. capillary malformation (*leptomeninges*)

Mosaic distribution [“pial”]

Choroidal plexus [gadolinium enhancement]

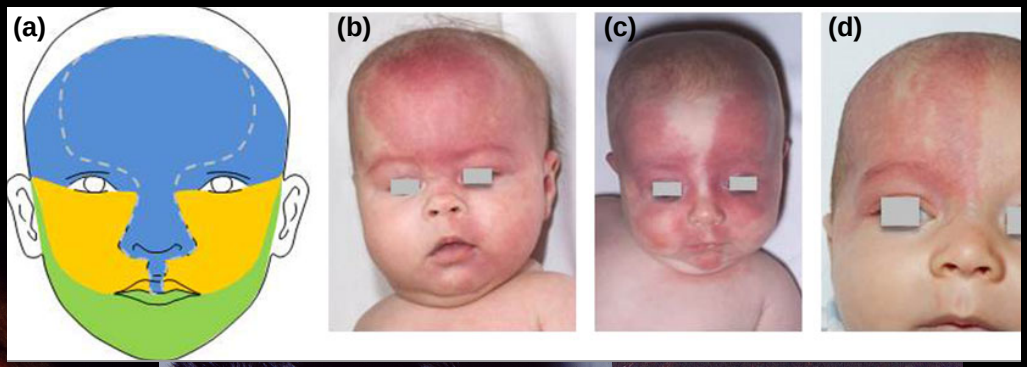
White matter anomalies

Vascular abnormalities

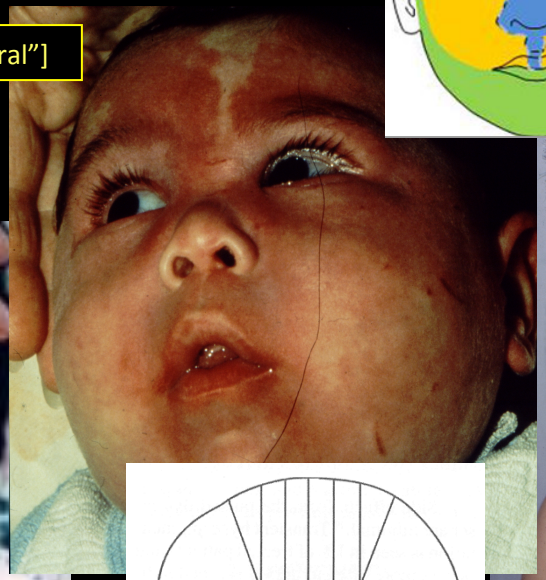
Gyral calcifications

Sturge-Weber syndrome

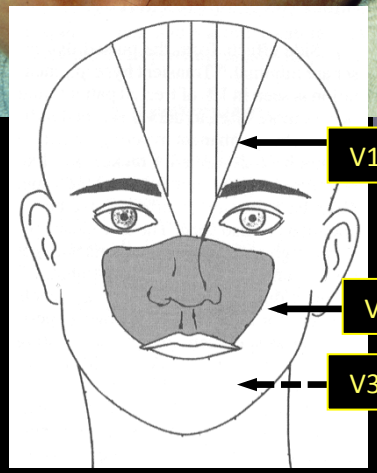
3. capillary malformation (cutaneous embryonic *microvasculature of the face*)



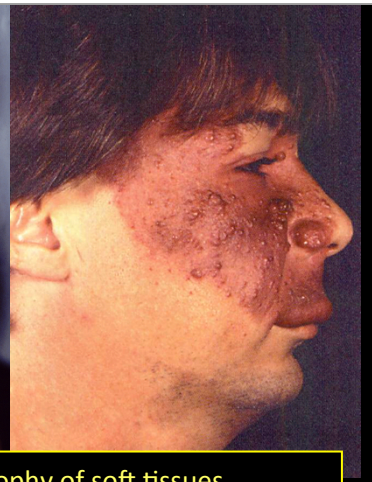
capillary malformation [“bilateral”]



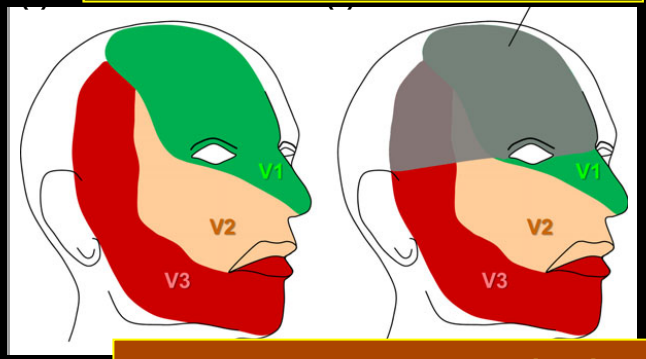
capillary malformation [“unilateral”]



Cranial nerve V [old]



Hypertrophy of soft tissues

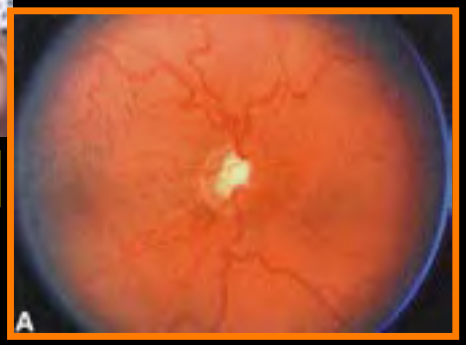


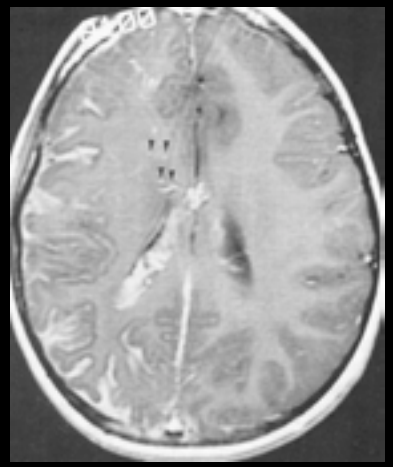
Facial embryonic vasculature [new]

Sturge-Weber syndrome

Phenotypical variability

Classical SWS [SWS type 1]





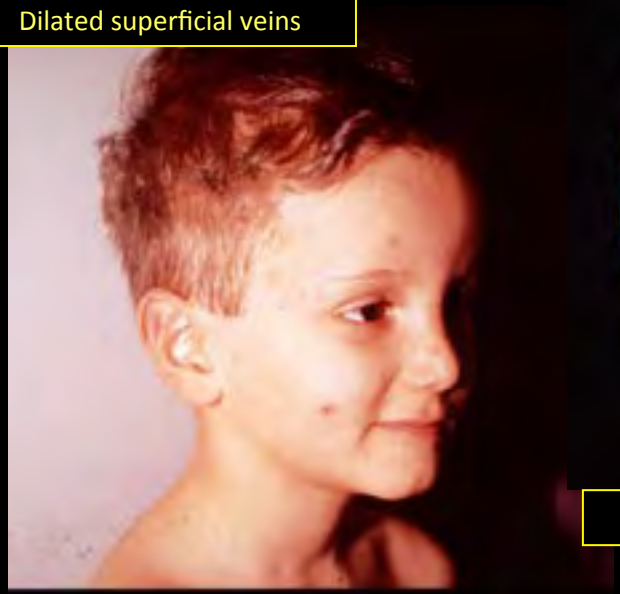
SWS without facial nevus [SWS type 3]

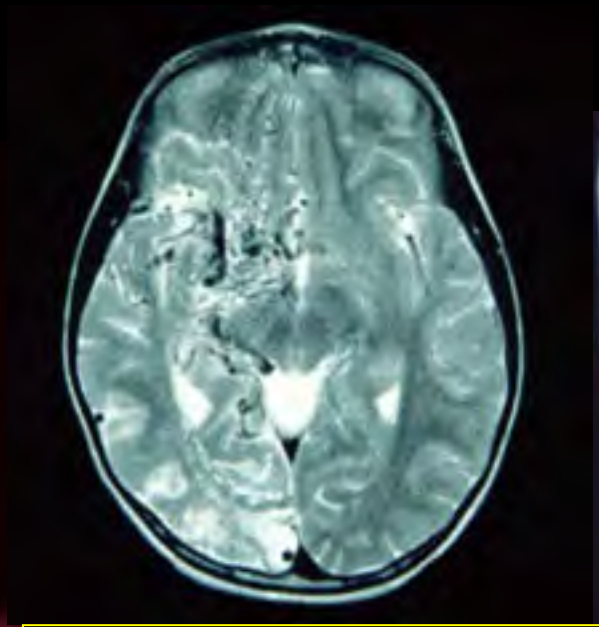
SWS with only facial nevus [SWS type 2]

1. Superficial & Deep venous abnormalities [*skin & brain*]

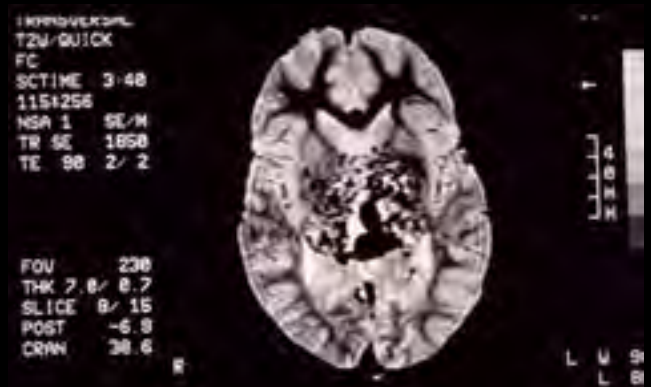
Wyburn-Mason syndrome

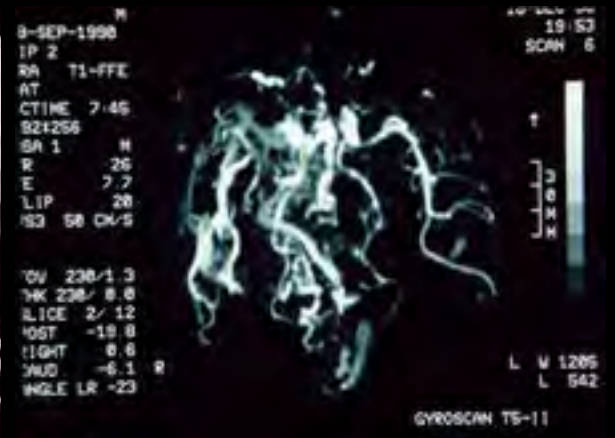
Dilated superficial veins





Thrombotic events - Infartcs - Ischemia

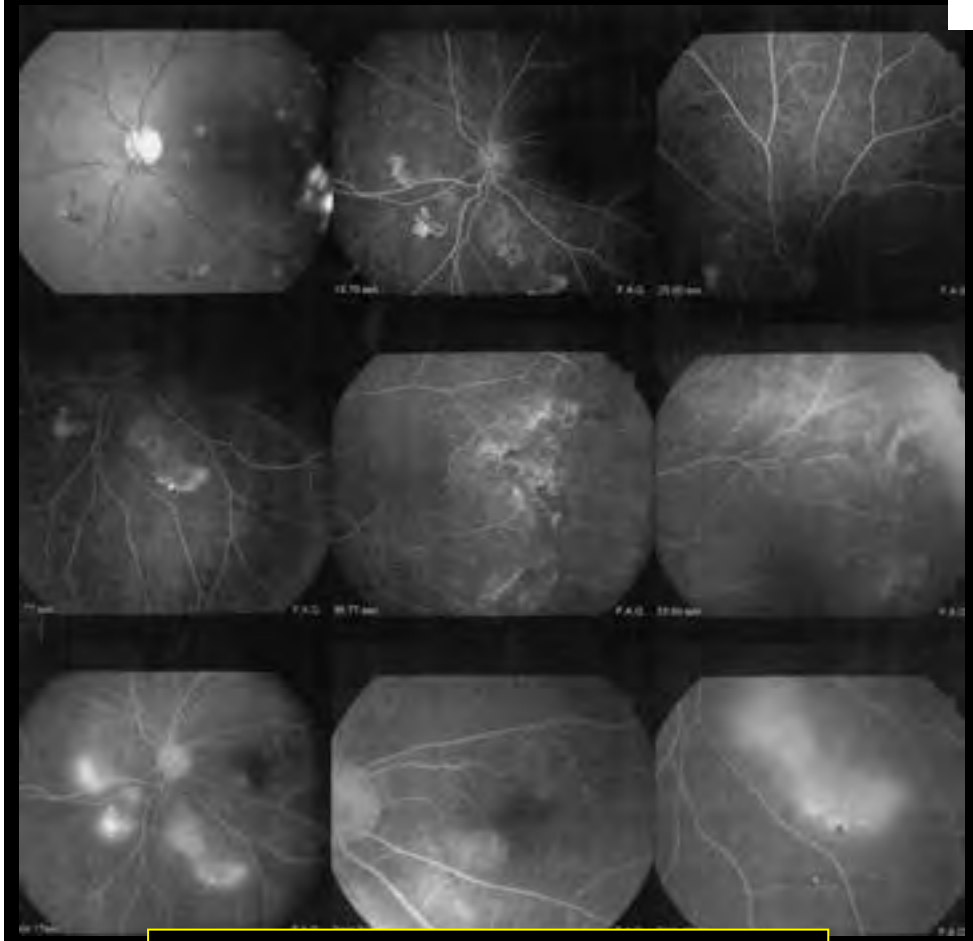




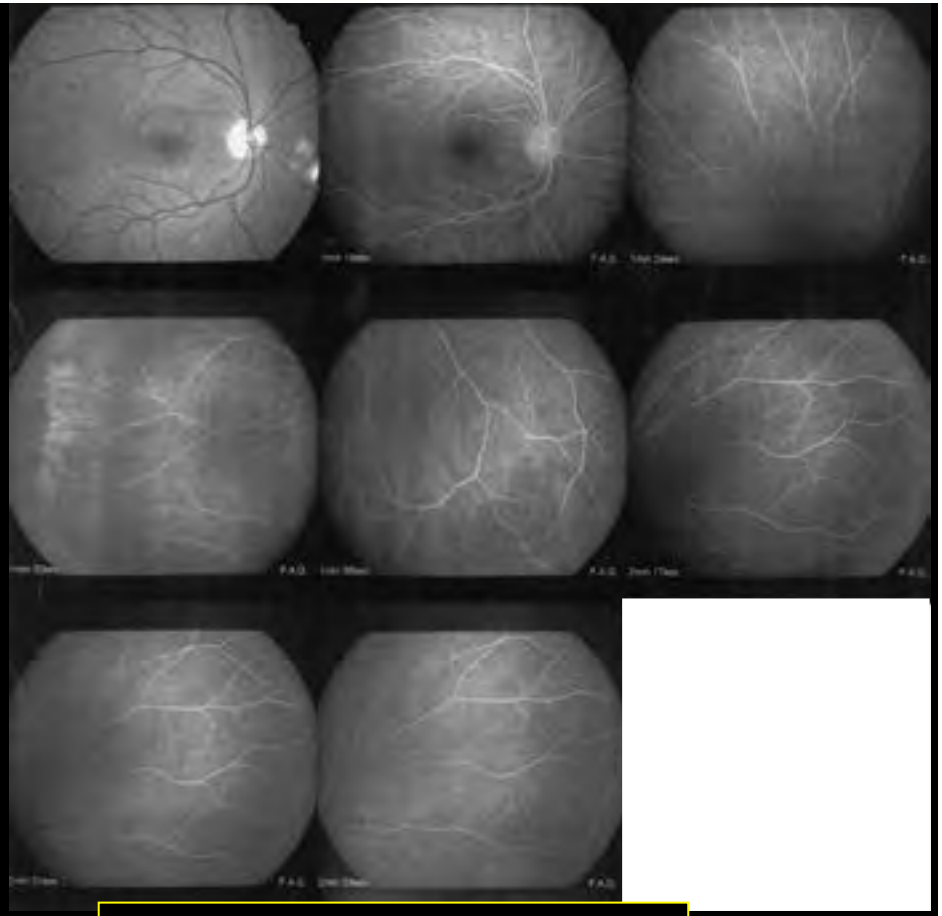
Dilatation of the deep venous vessels

Wyburn-Mason syndrome

1. Venous abnormalities [*retinal*]



Dilatation of the venous vessels [retinal]



Normal contralateral fundoscopy

Mixed vascular nevus syndrome

1. Paired vascular nevi

Nevus anemicus

Nevus telangiectaticus

Nevus anemicus

Nevus telangiectaticus

1. Paired vascular nevi

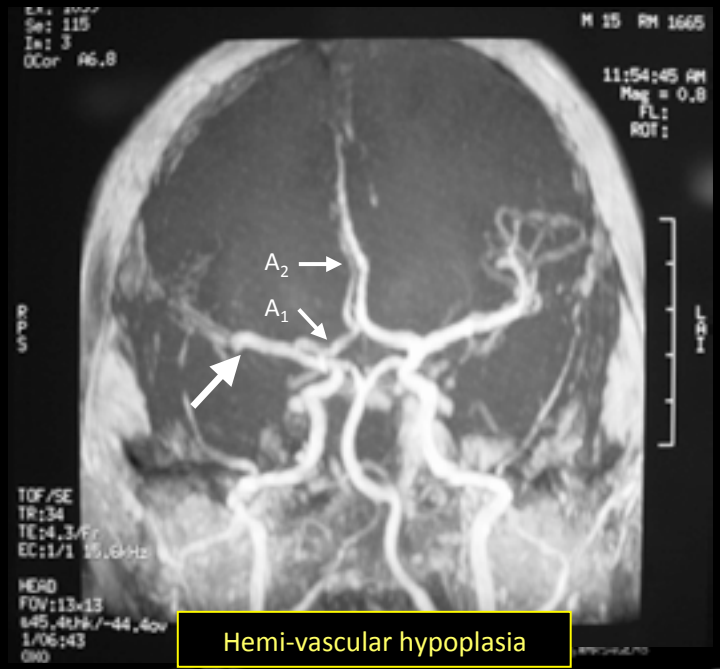
Mixed vascular nevus syndrome

Nevus telangiectaticus

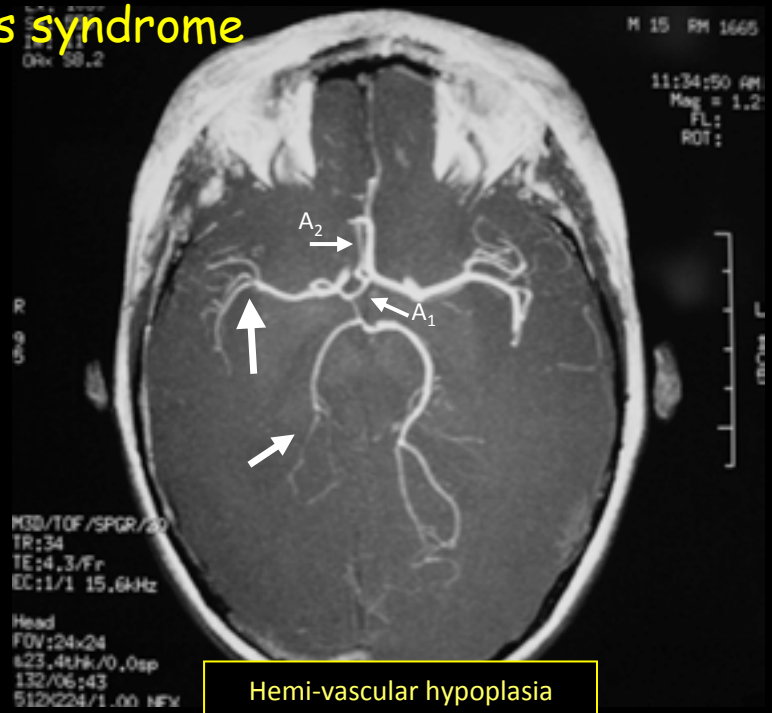
Nevus anemicus

Mixed vascular nevus syndrome

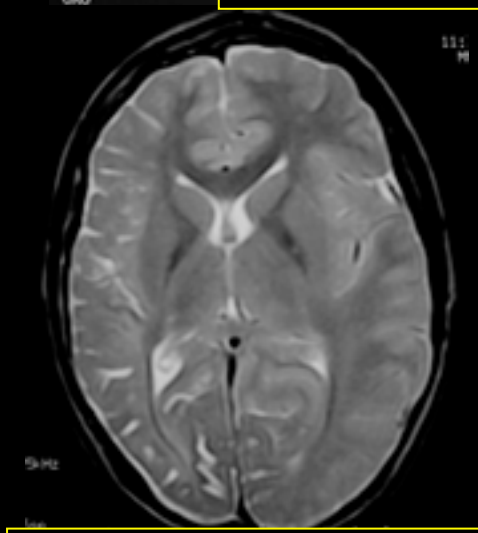
2. Dyke-Davidoff-Masson syndrome



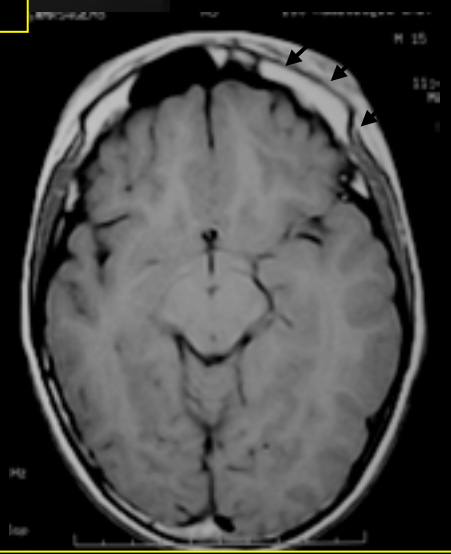
Hemi-vascular hypoplasia



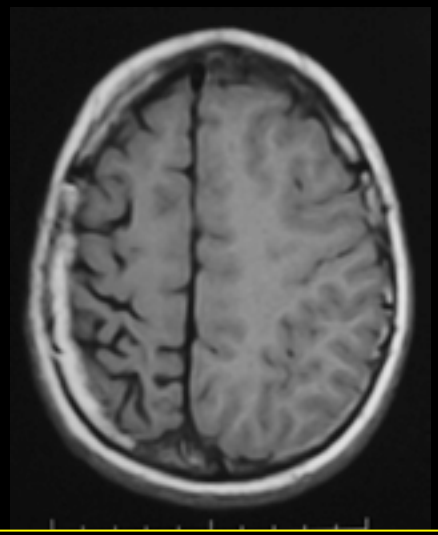
Hemi-vascular hypoplasia



unilateral decreased cerebral growth

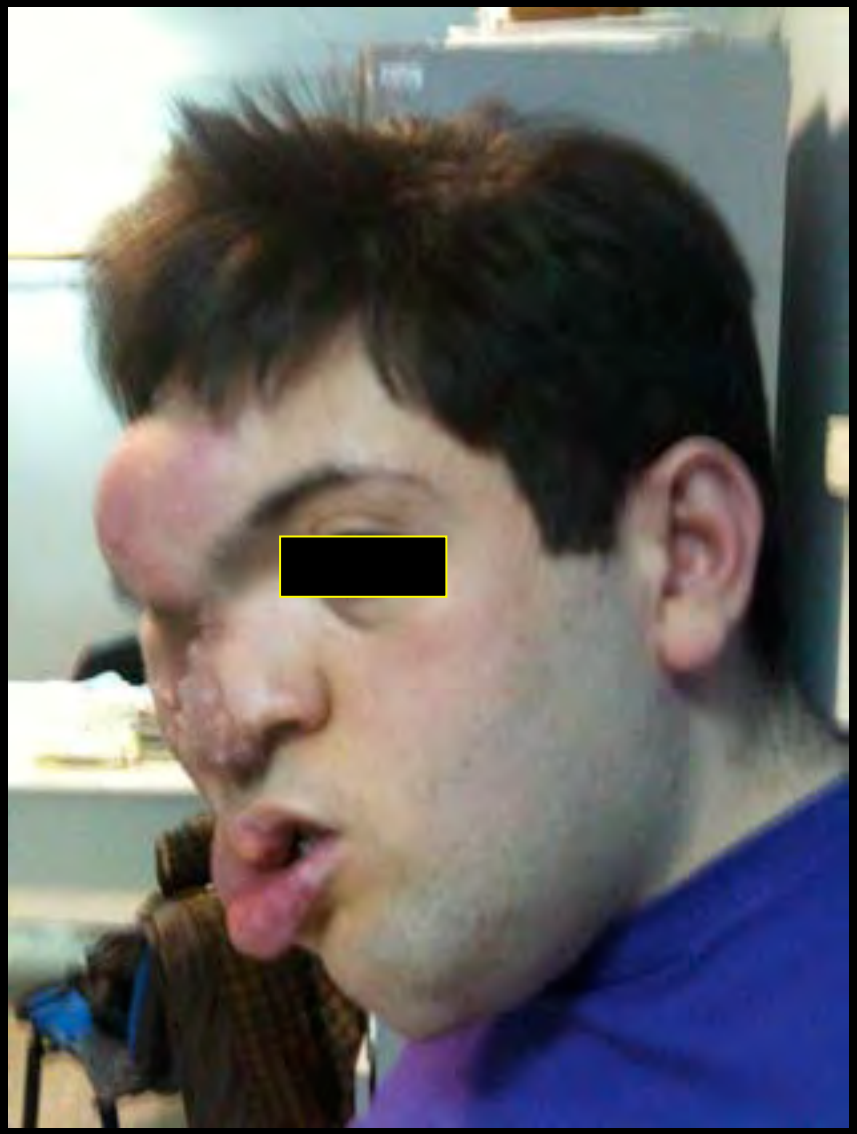


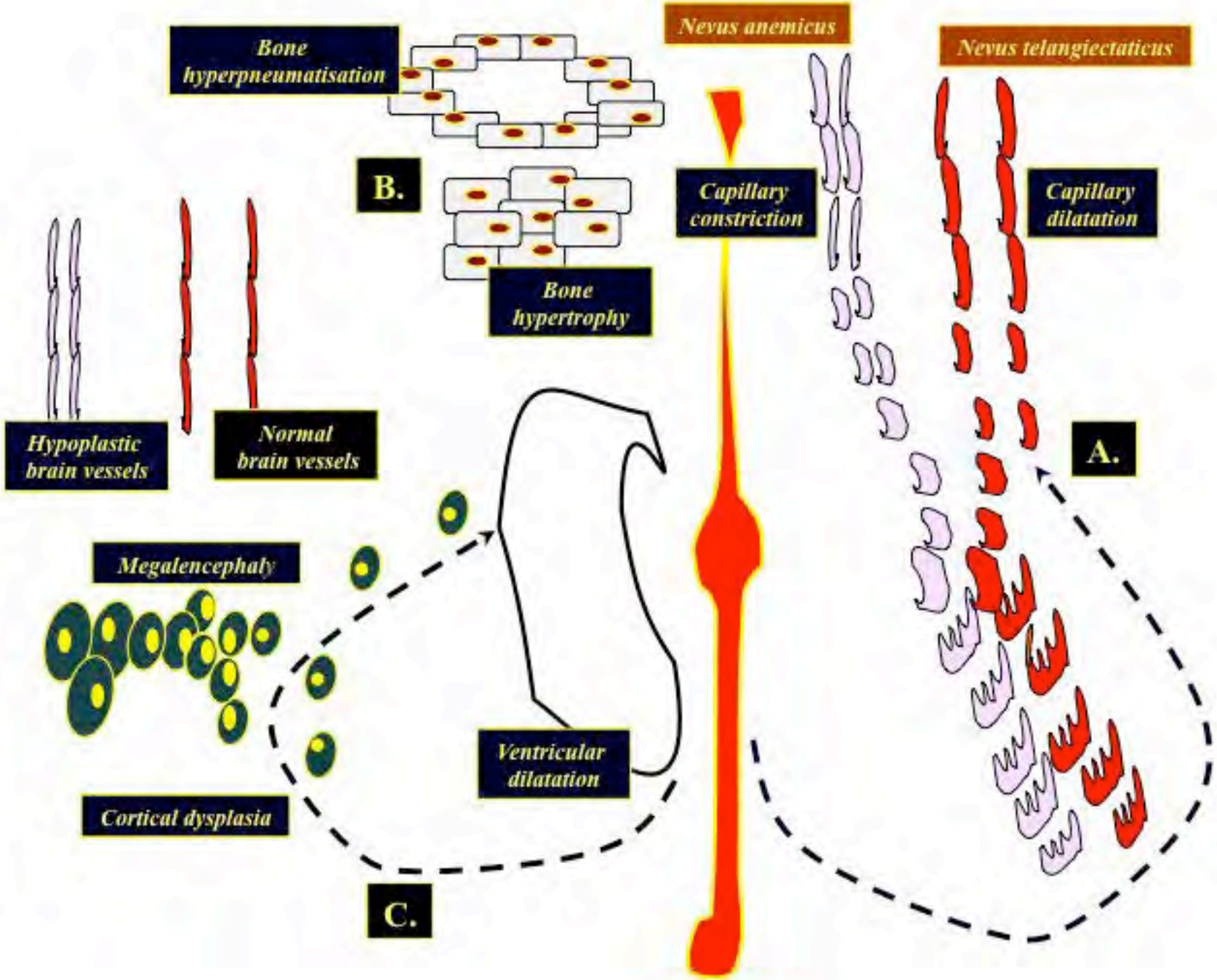
Sinuses hyperpneumatisation



unilateral decreased cerebral growth

Mixed vascular nevus syndrome

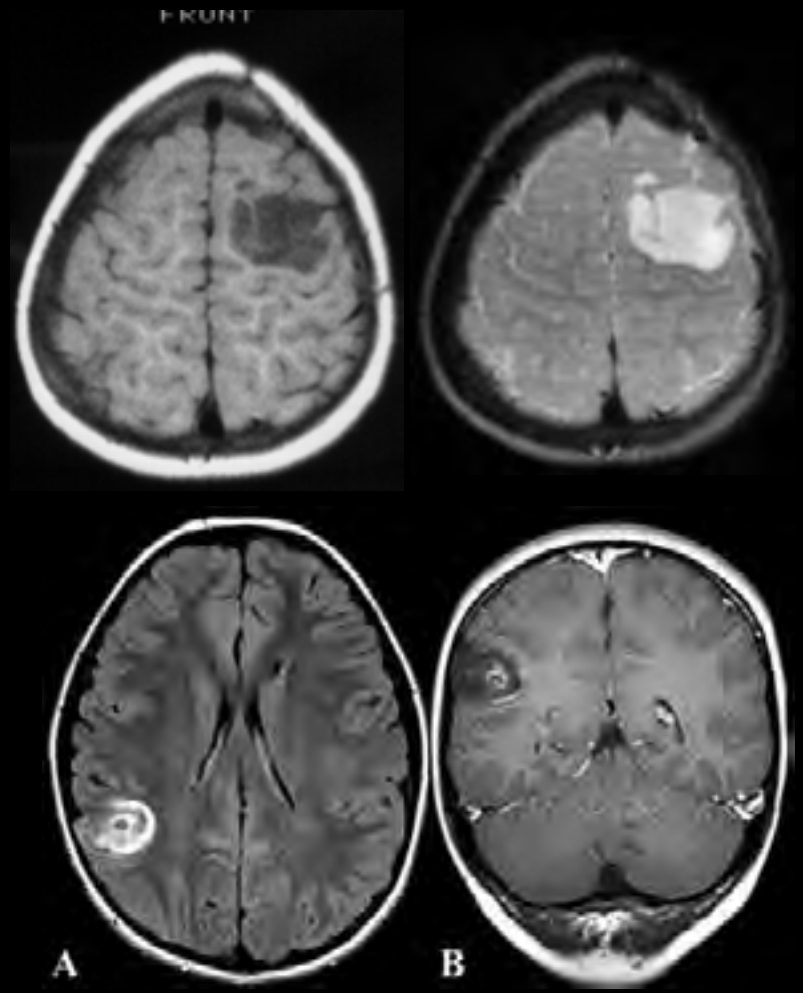


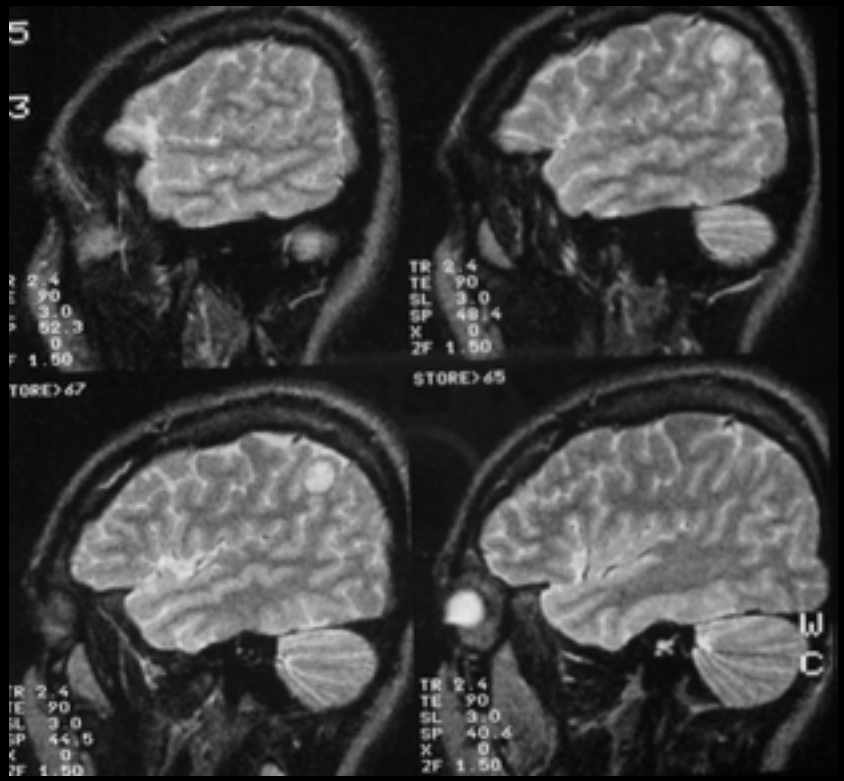

Nevus anemicus
Nevus telangiectaticus
Bone hyperpneumatisation
B.
Capillary constriction
Capillary dilatation
Bone hypertrophy
Hypoplastic brain vessels
Normal brain vessels
A.
Megalencephaly
Ventricular dilatation
Cortical dysplasia
C.

MALFORMAZIONI dello SVILUPPO CORTICALE

I. MALFORMAZIONI DA ANOMALIE DELLA PROLIFERAZIONE/APOPTOSI DEI PRECURSORI NEURONALI E GLIALI

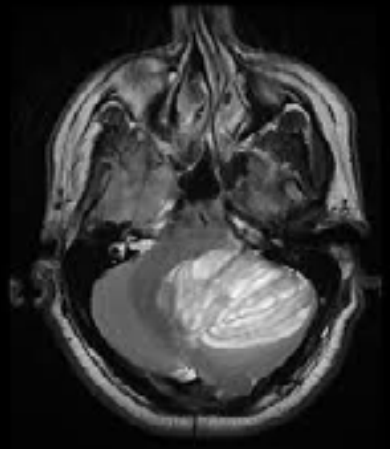
A) PROLIFERAZIONE ANOMALA (TIPI CELLULARI ANOMALI)

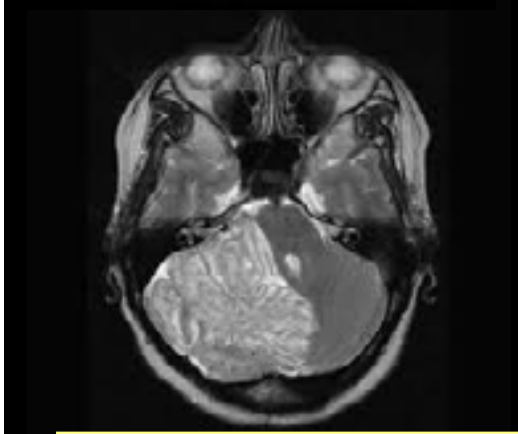


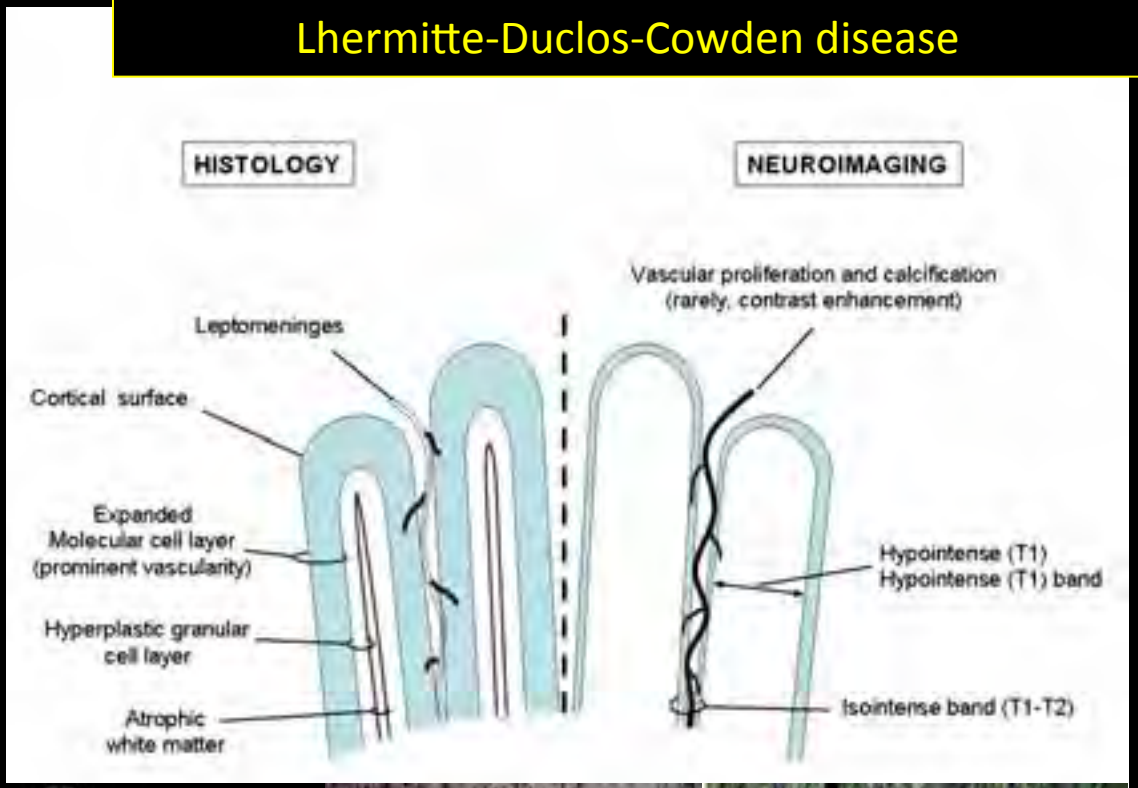


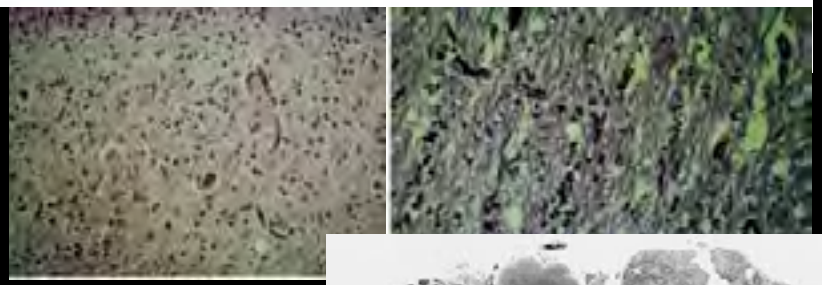
Lhermitte-Duclos-Cowden disease

Jean Jacques LHERMITTE
[1877 - 1959]



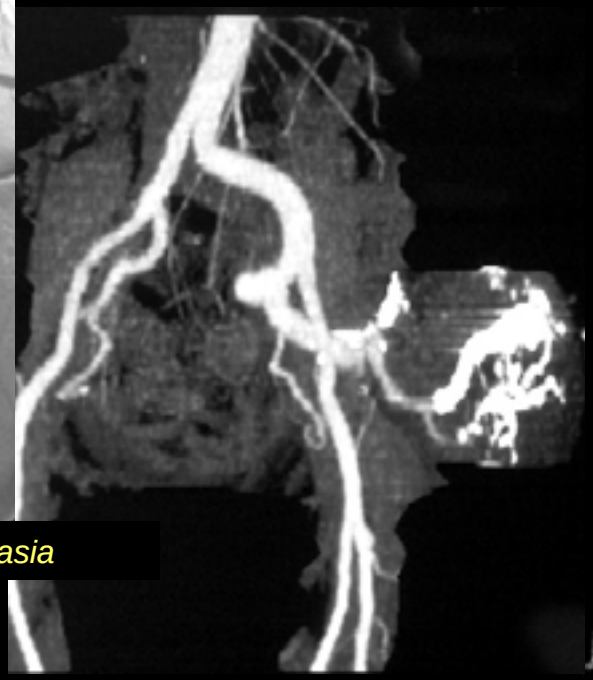




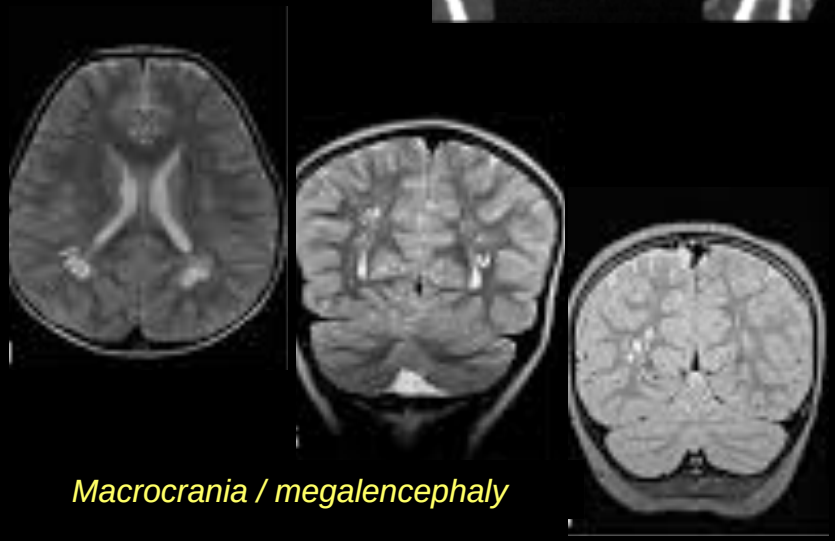


Lhermitte JJ, Duclos P. Sur un ganglioneurome diffuse du cortex du cervelet. *Bulletin de l'Association Francaise pour l'etude du Cancer*. 1920;9:99-107.

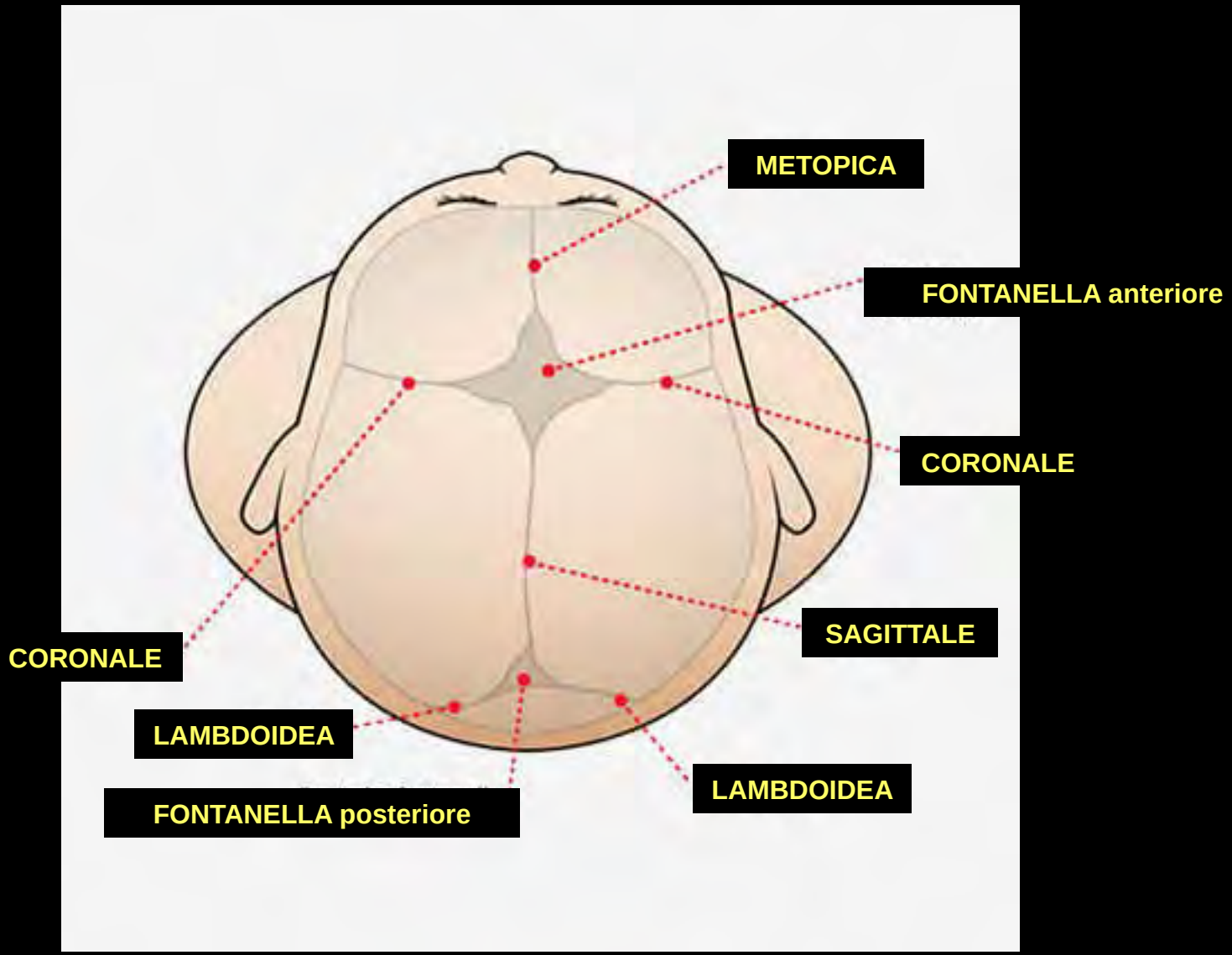
Lhermitte-Duclos-Cowden disease

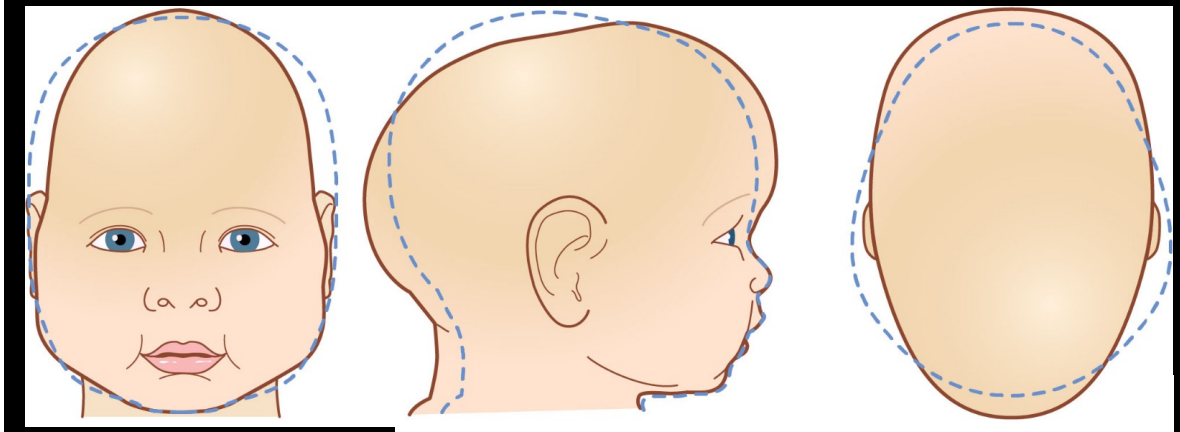


Vascular venous dysplasia

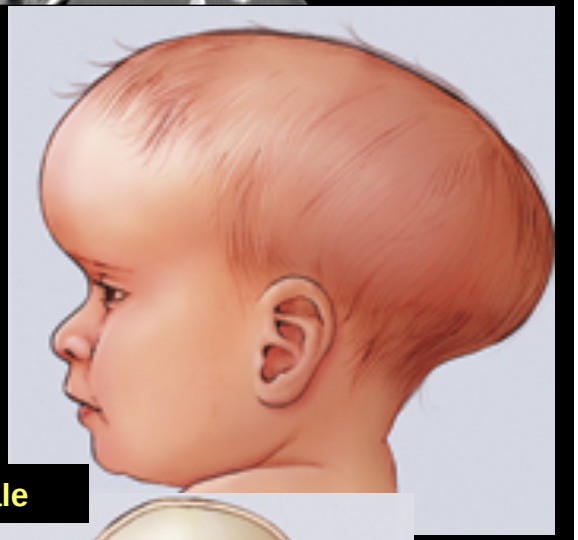


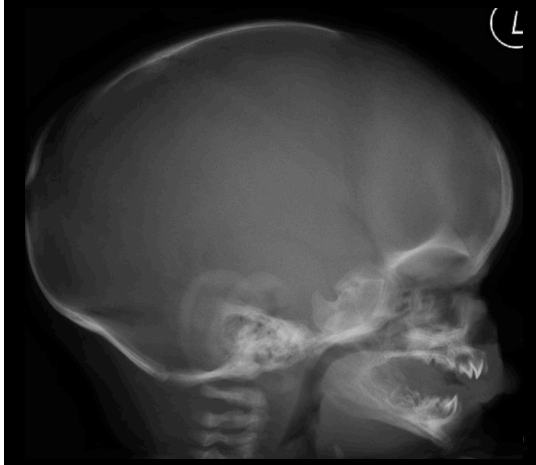
Macrocrania / megalencephaly


METOPICA
FONTANELLA anteriore
CORONALE
SAGITTALE
CORONALE
LAMBDOIDEA
LAMBDOIDEA
FONTANELLA posteriore

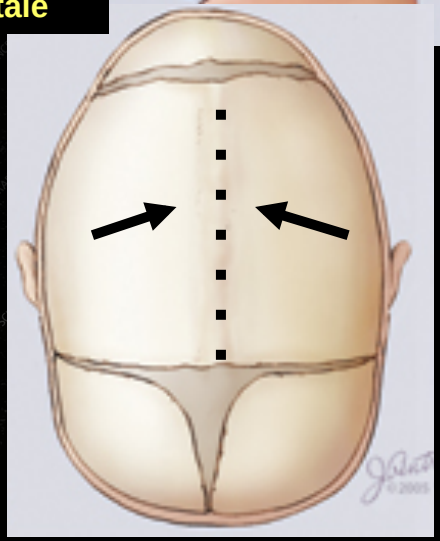


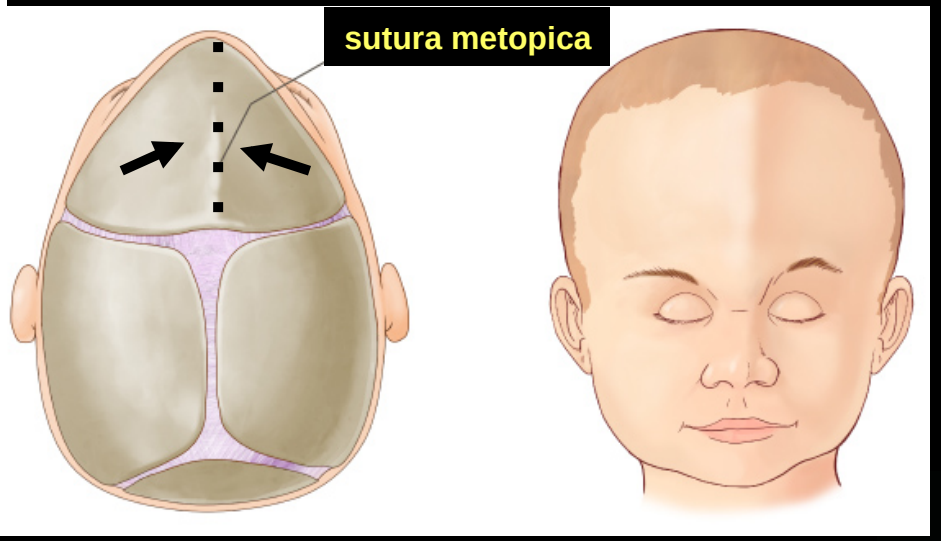
SCAFOCEFALIA



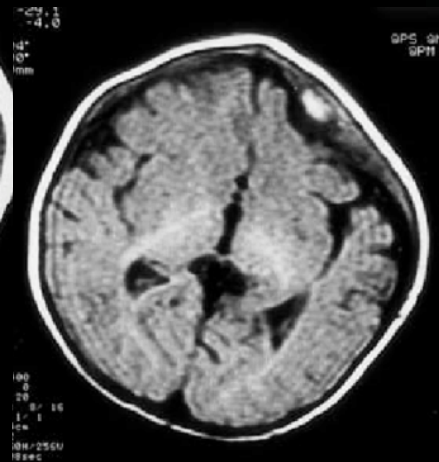


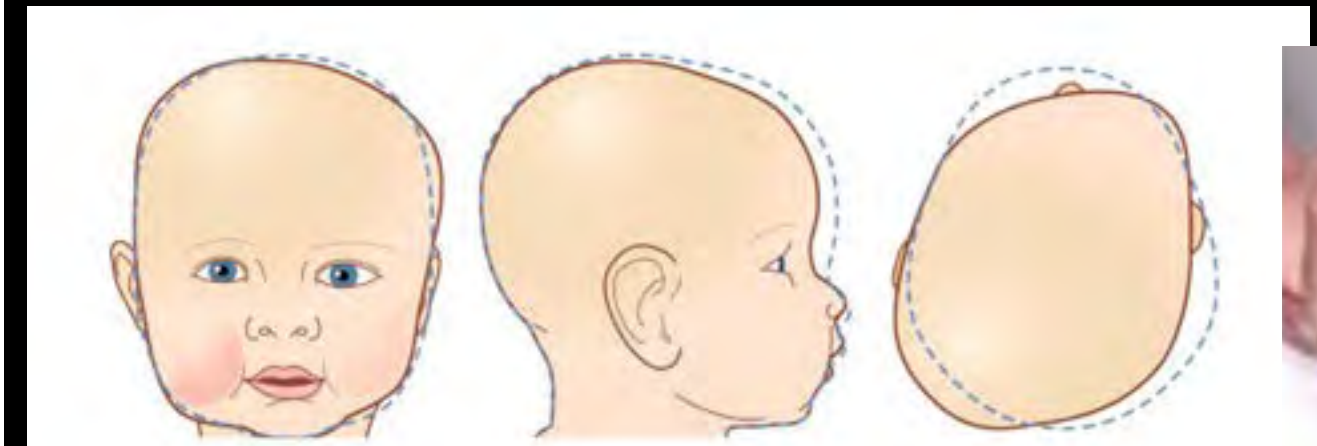
sutura sagittale



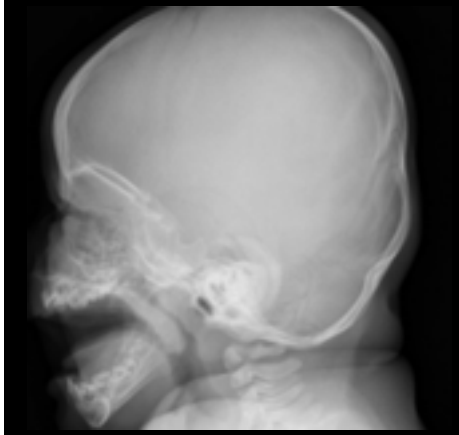


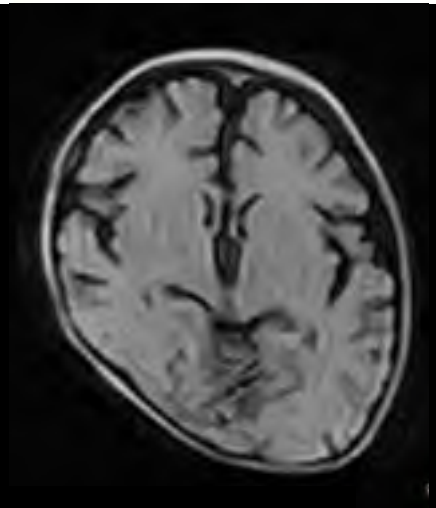
TRIGONOCEFALIA



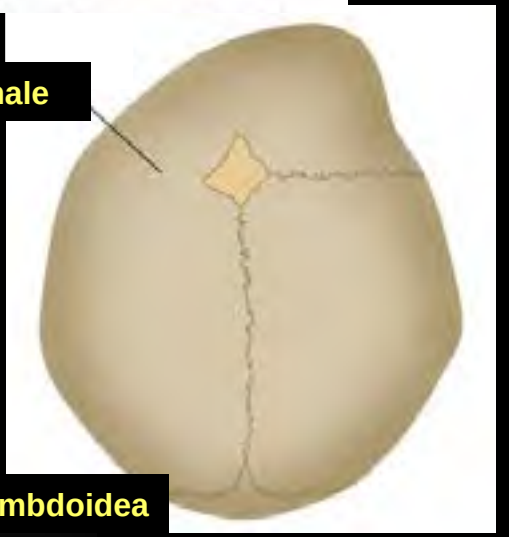


PLAGIOCEFALIA

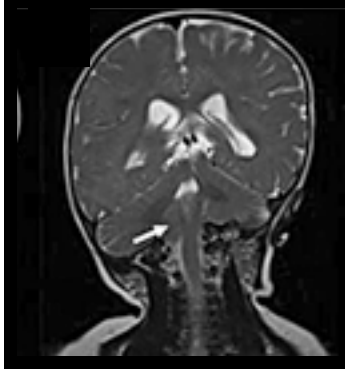


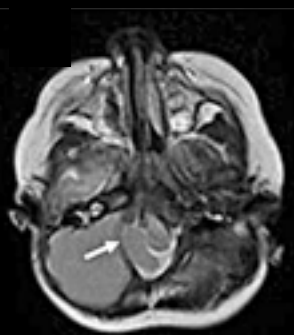


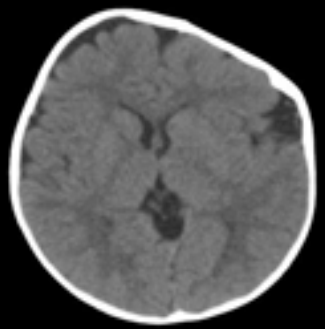
sutura coronale

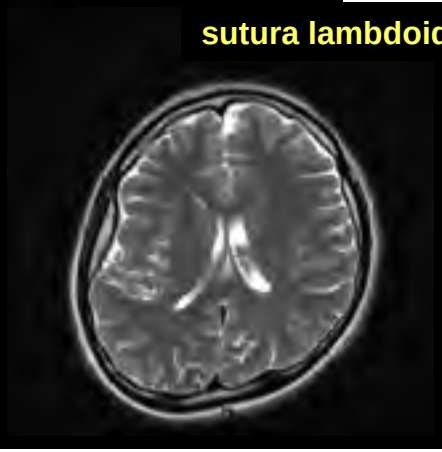


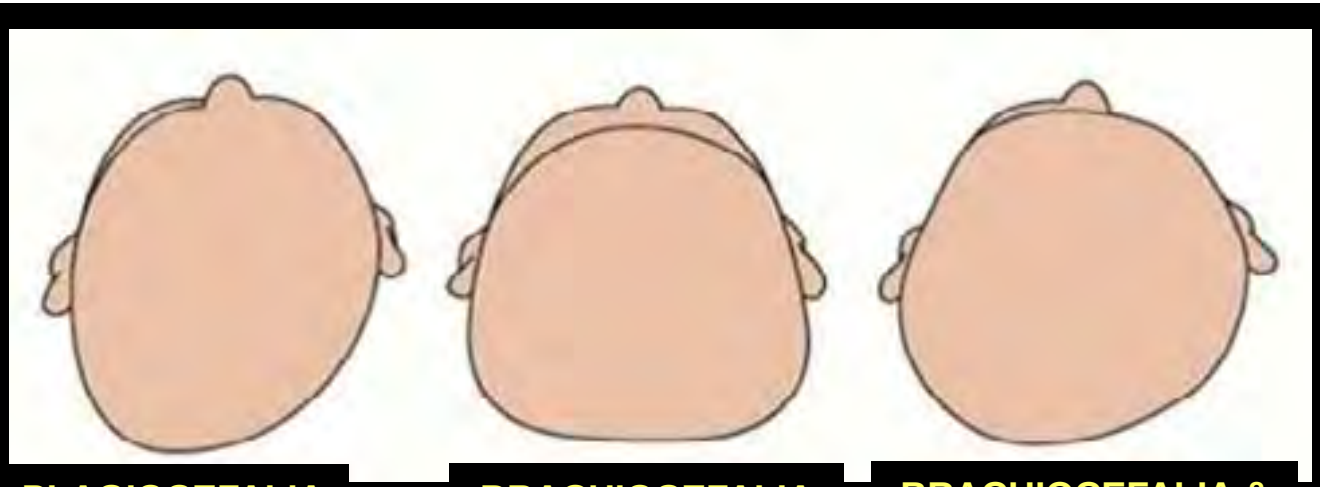
sutura lambdoidea







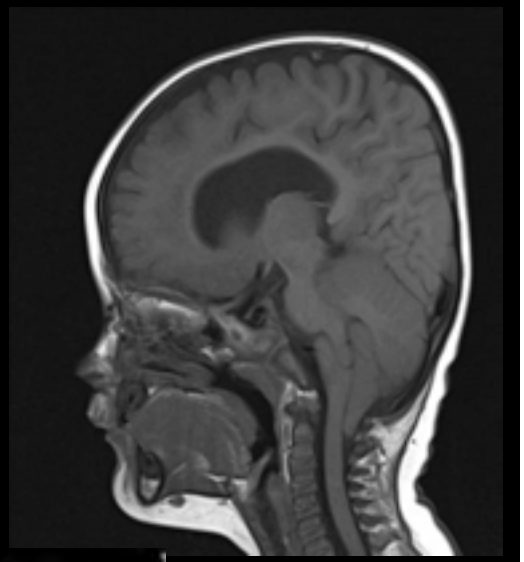


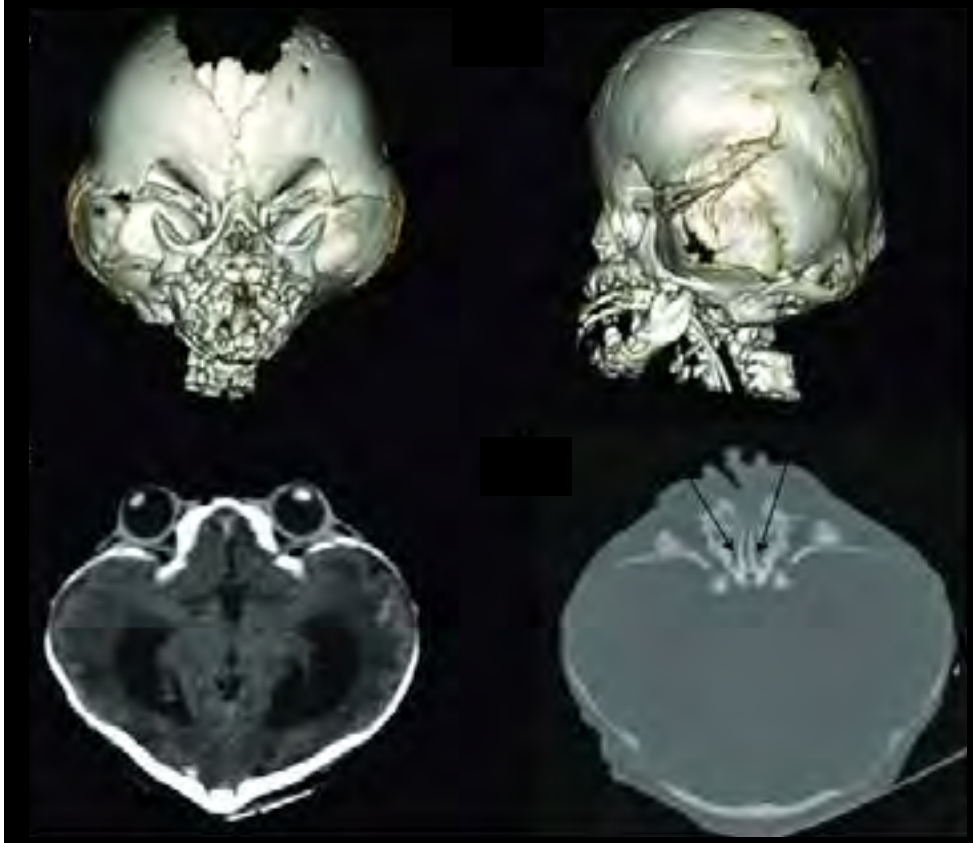


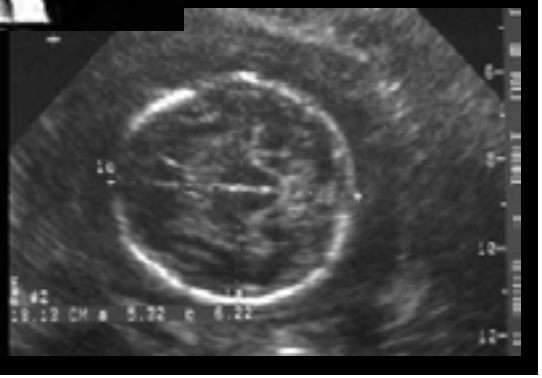
PLAGIOCEFALIA

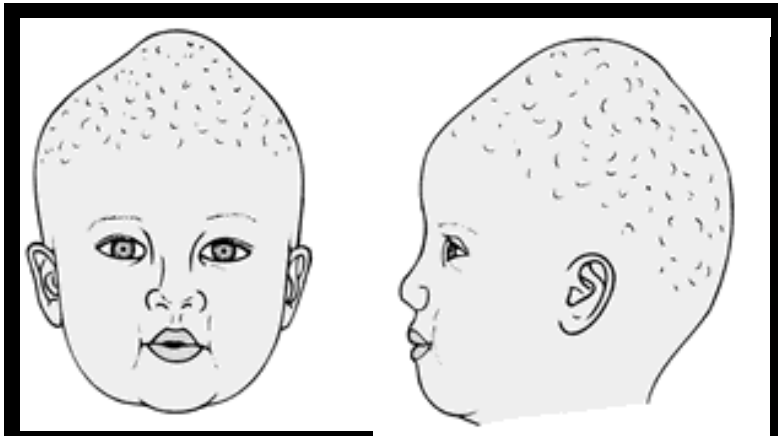
BRACHIOCEFALIA

BRACHIOCEFALIA & PLAGIOCEFALIA

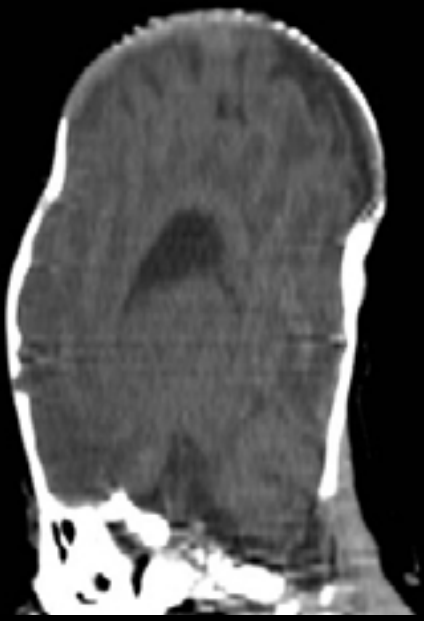


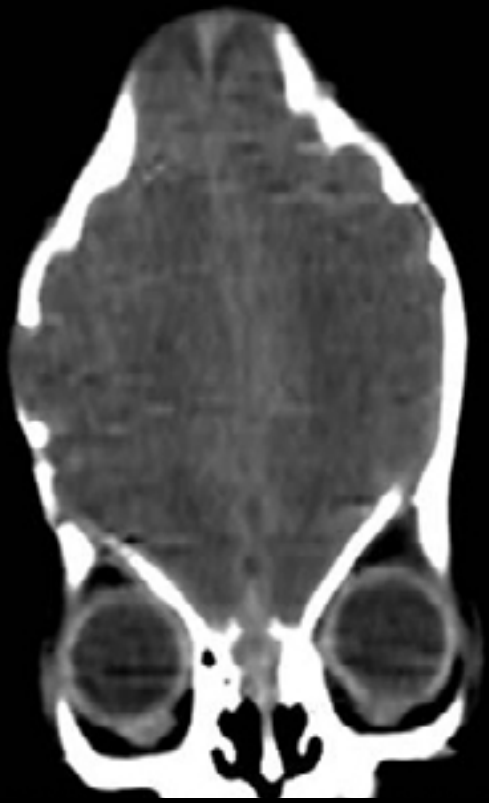


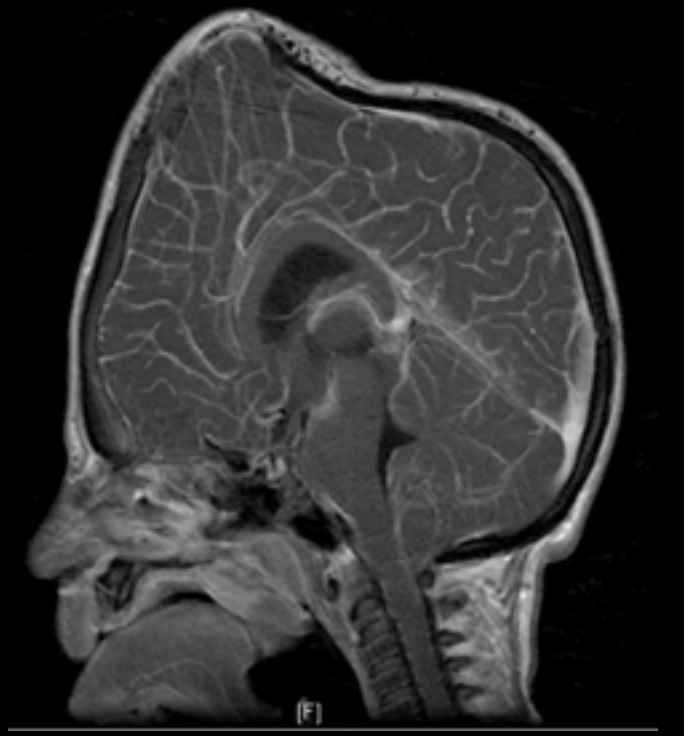




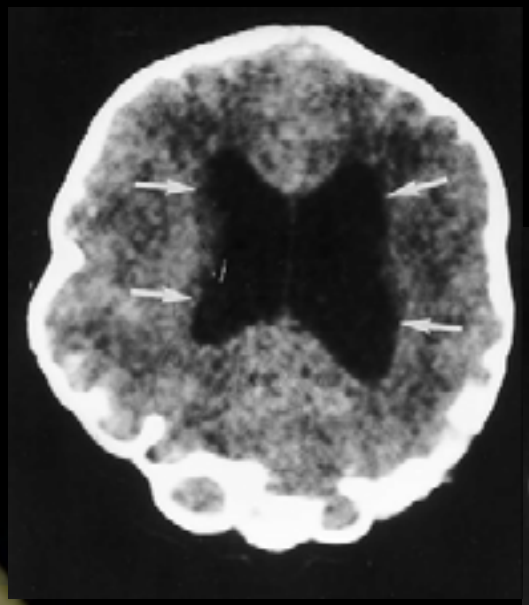
OSSICEFALIA

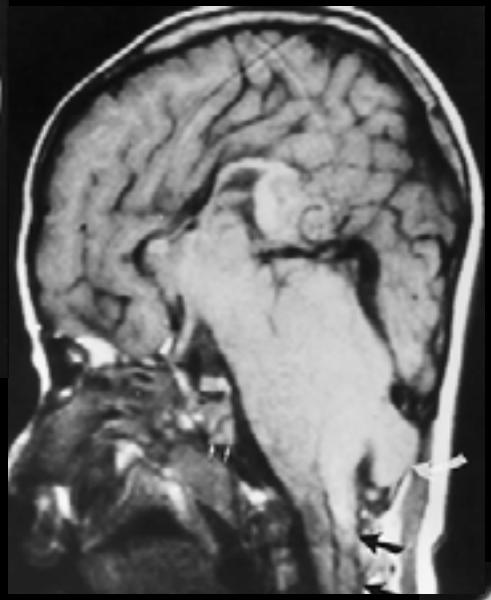


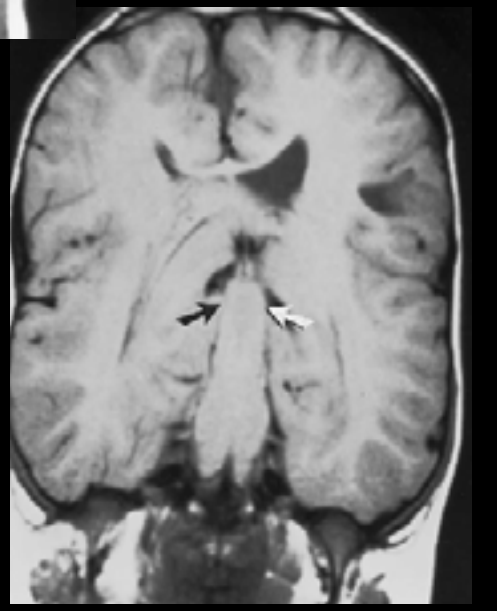


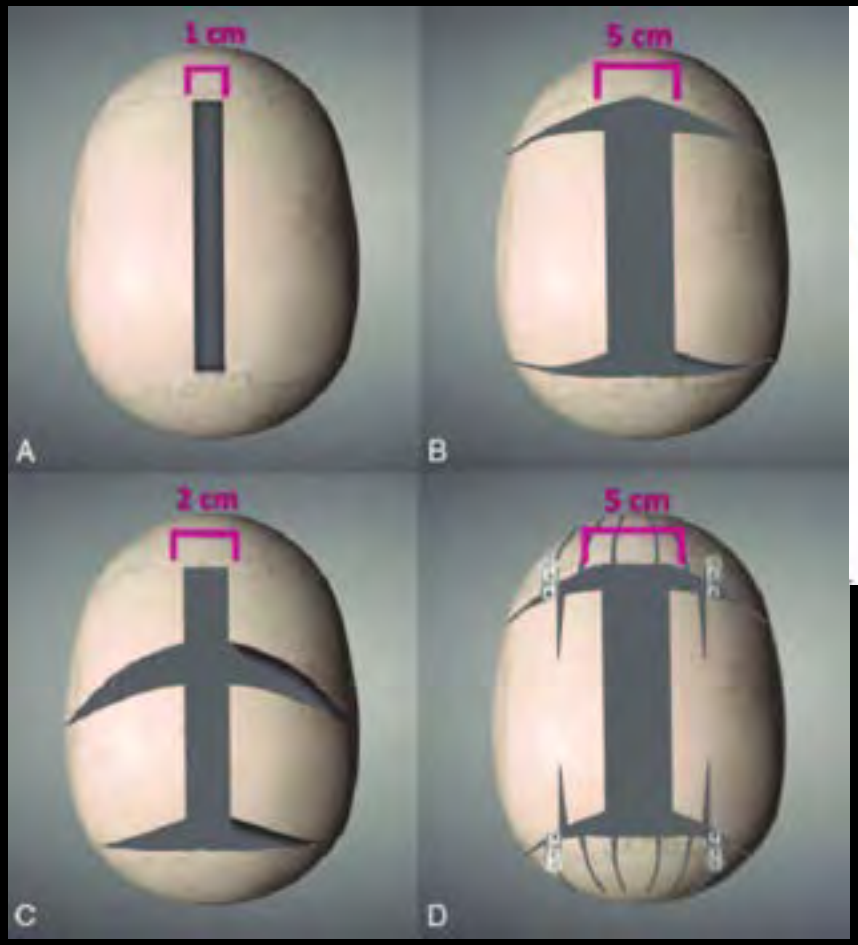


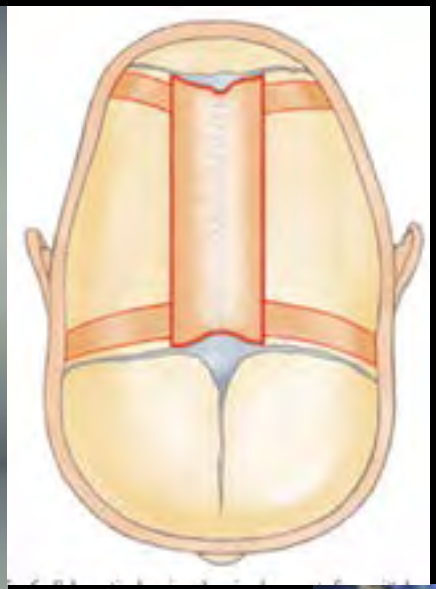
PANSINOSTOSI

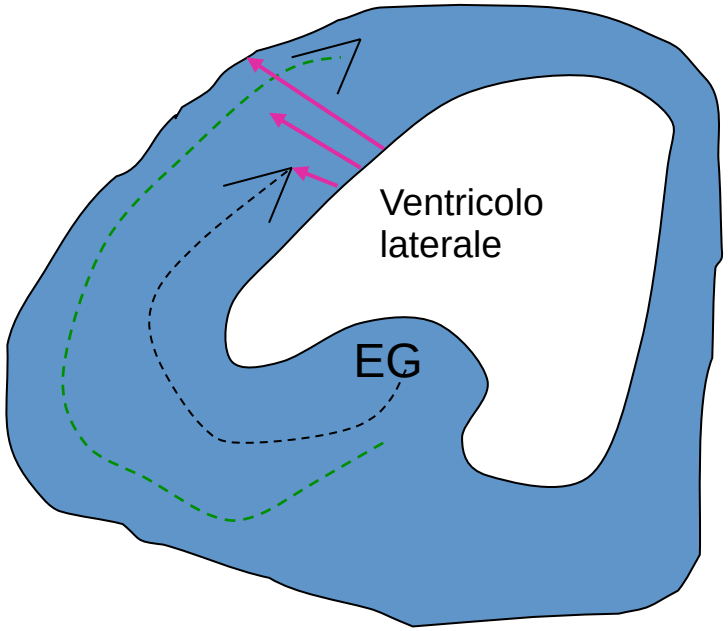


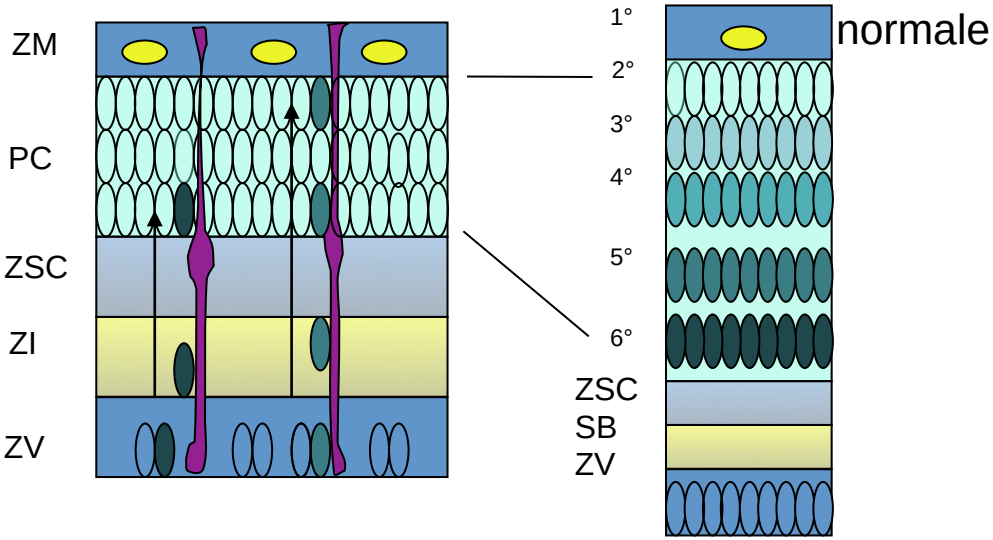




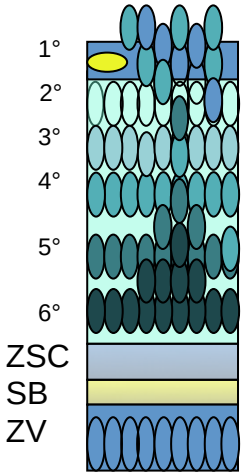

1 cm
5 cm
A
B
2 cm
5 cm
C
D



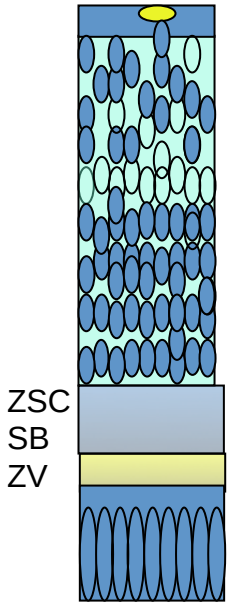




Complesso
cobblestone



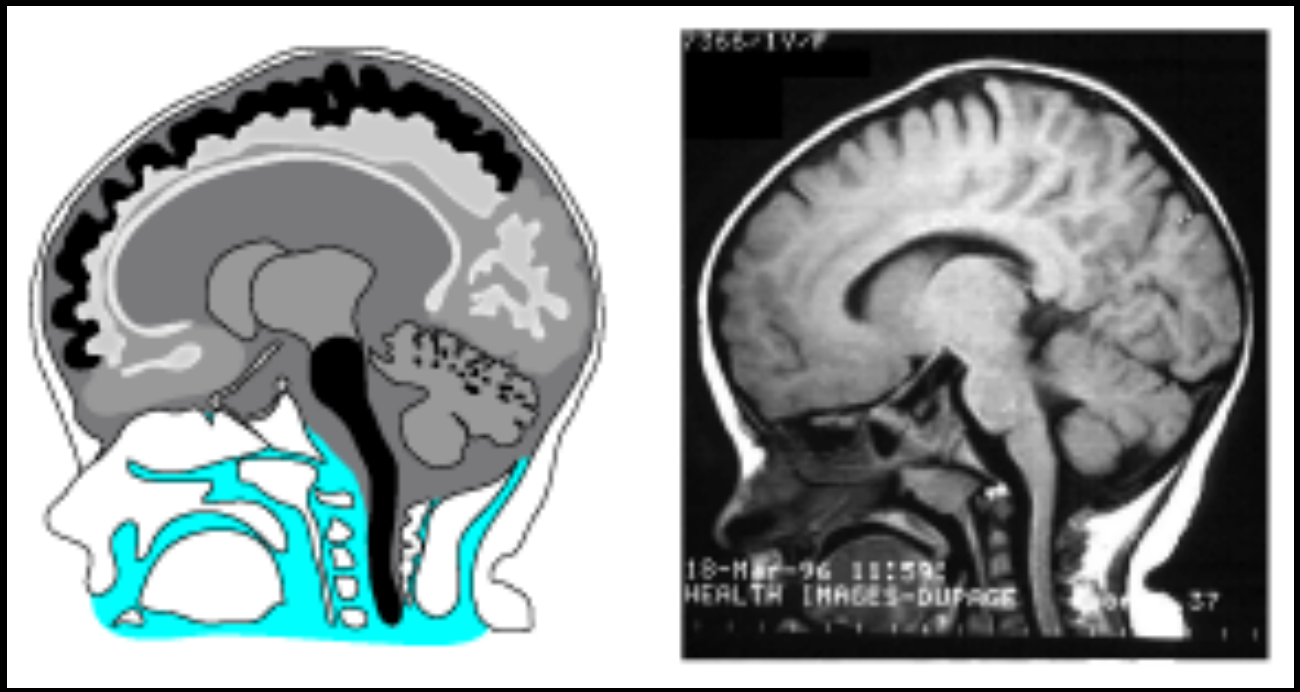
Lissencefalia

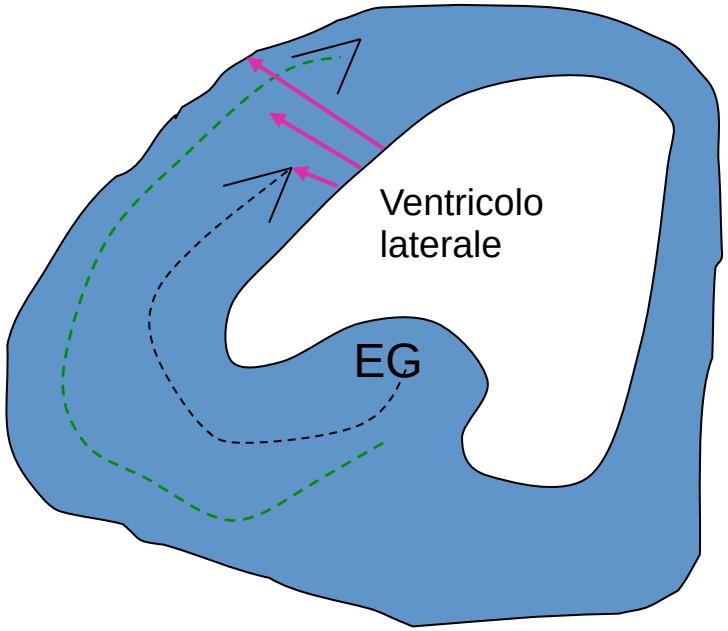


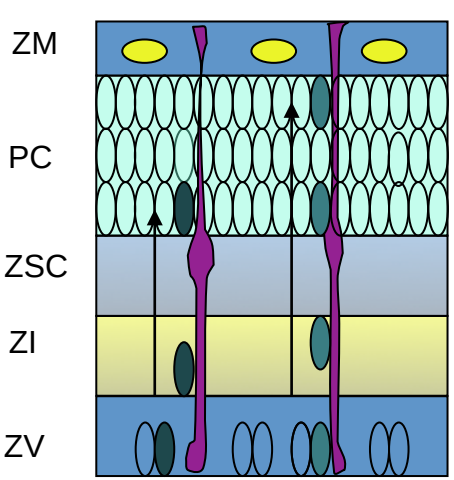
MALFORMAZIONI dello SVILUPPO CORTICALE

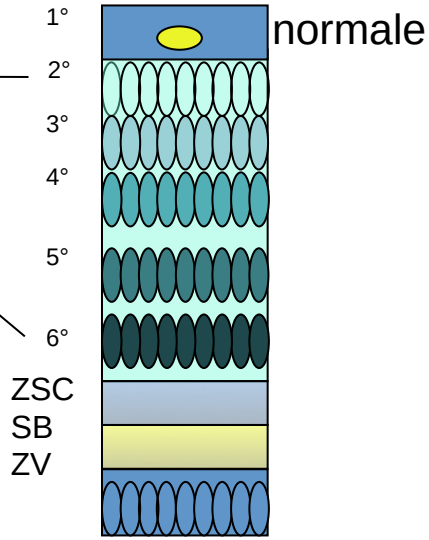
II. MALFORMAZIONI DA ANOMALIE DELLA MIGRAZIONE NEURONALE

B) COMPLESSO COBBLESTONE



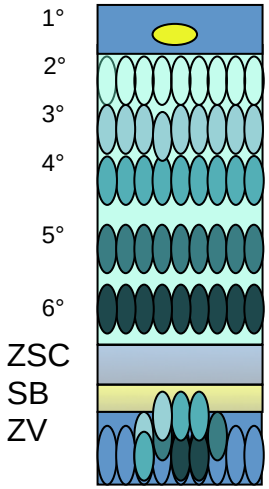




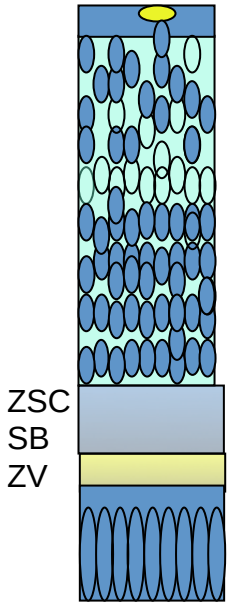


normale

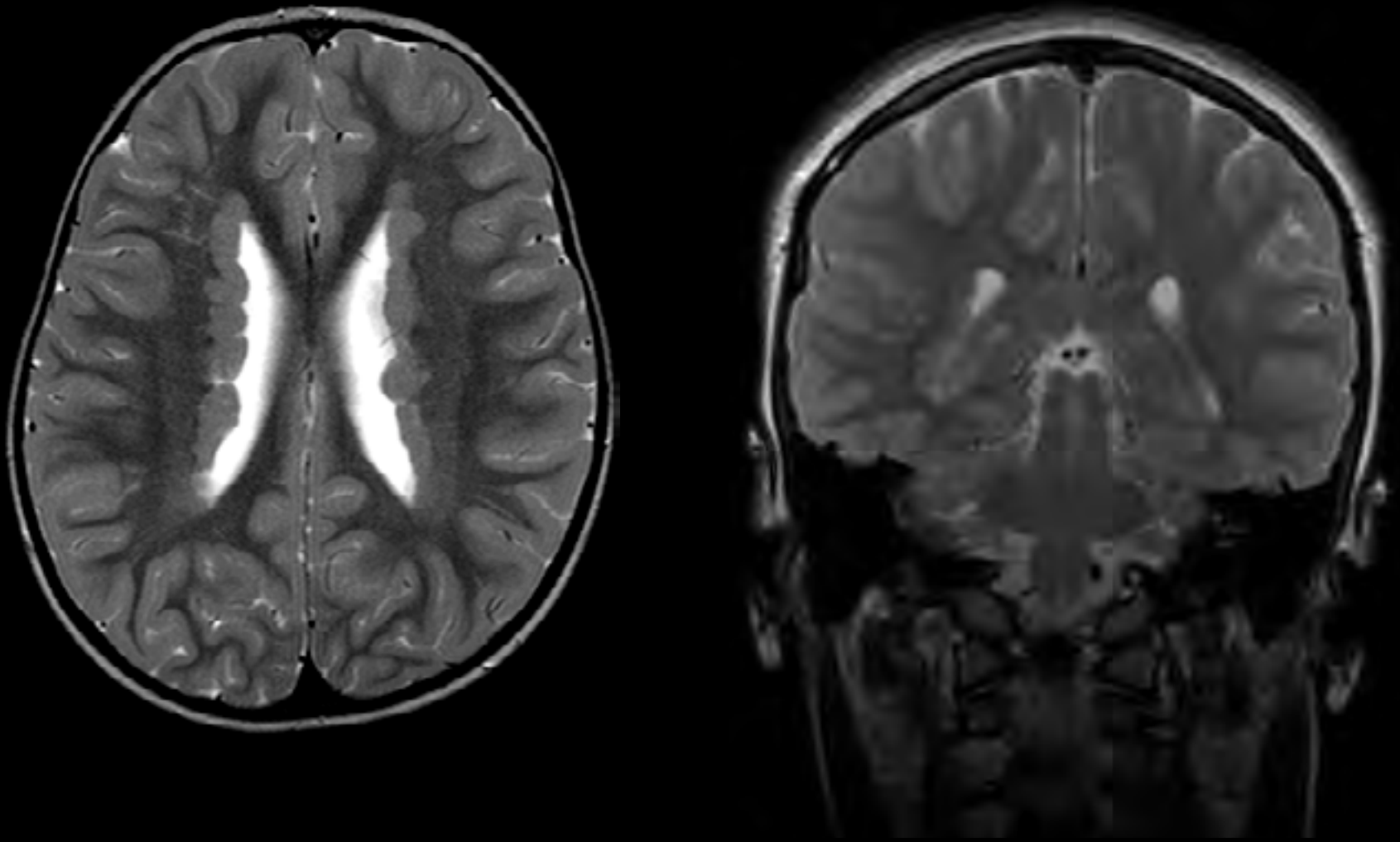
Eterotopie periventricolari



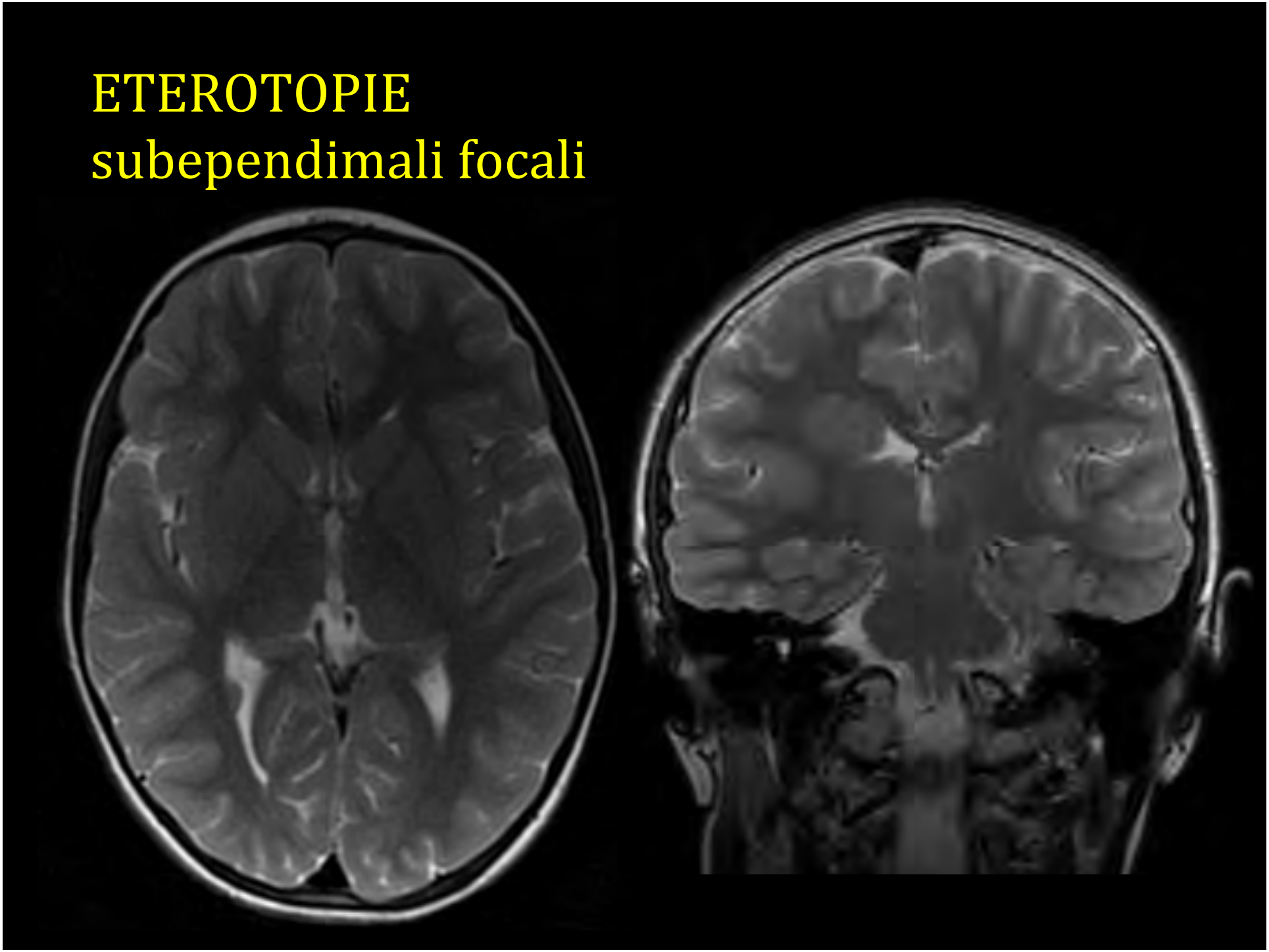
Lissencefalia



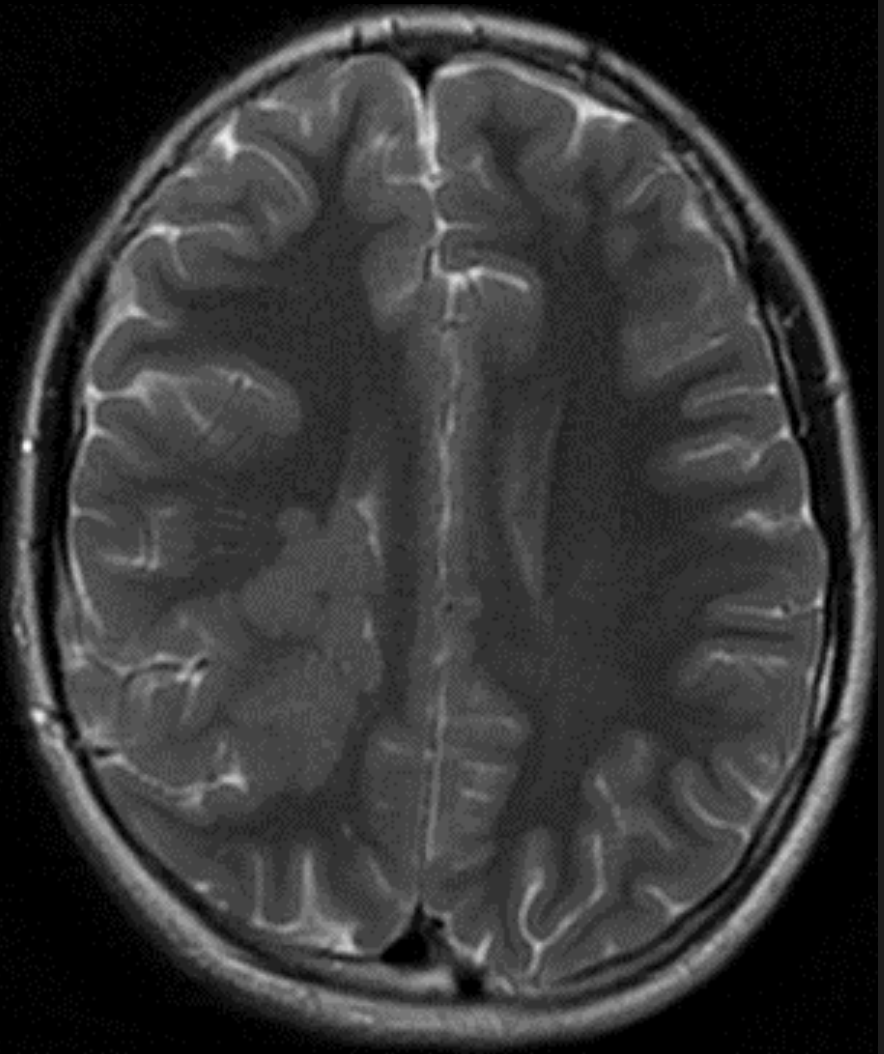
ETEROTOPIE
subependimali diffuse (BPNH)

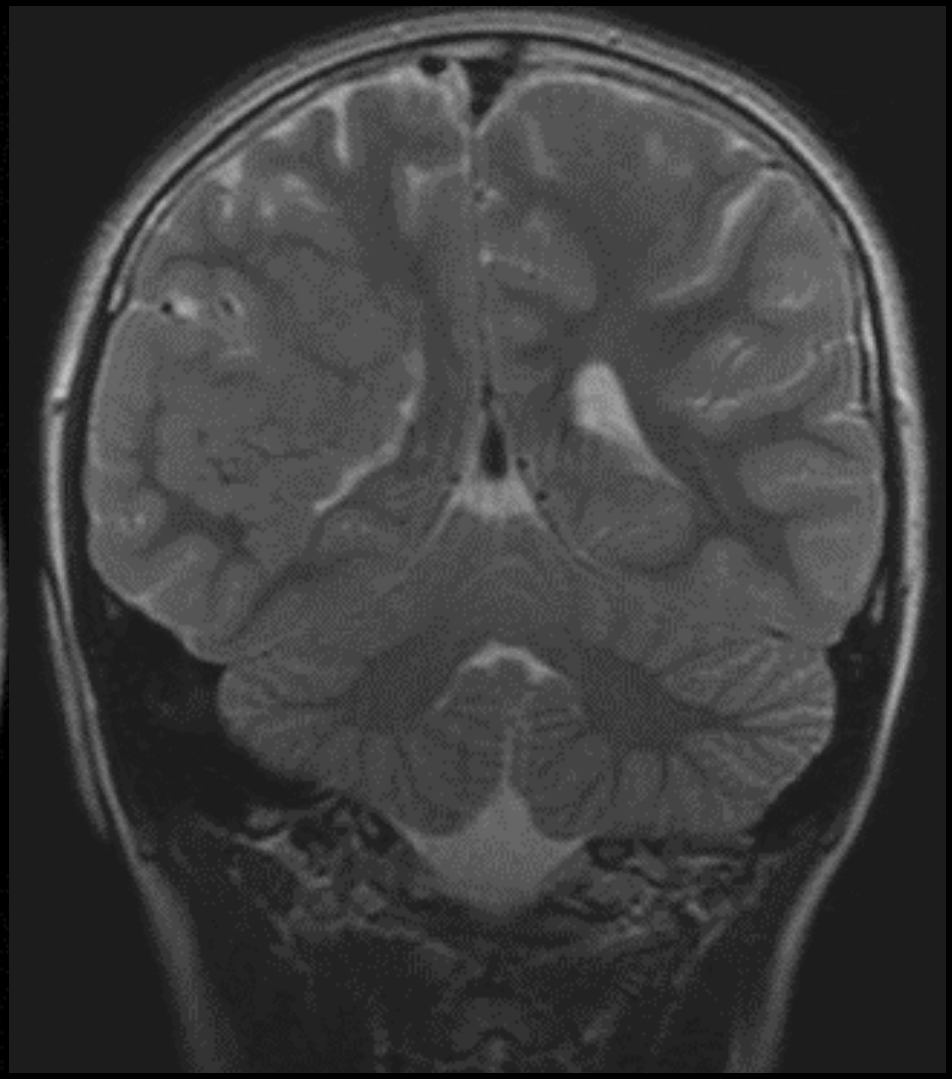


ETEROTOPIE
subependimali focali

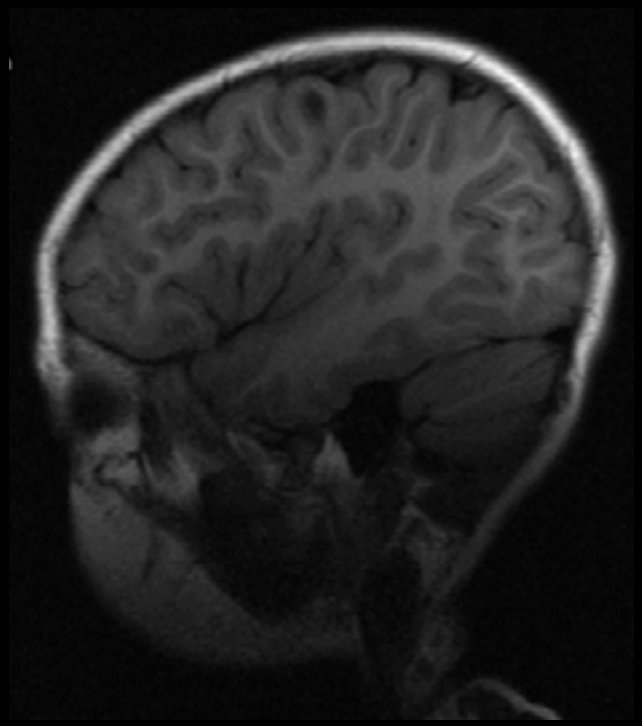


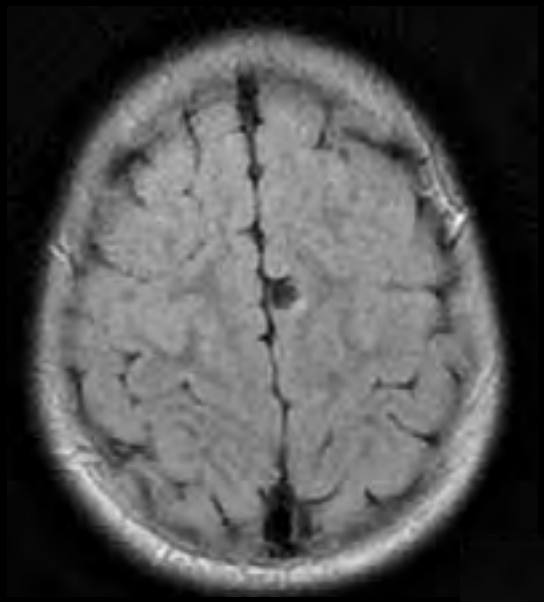
Eterotopie subependimali estese

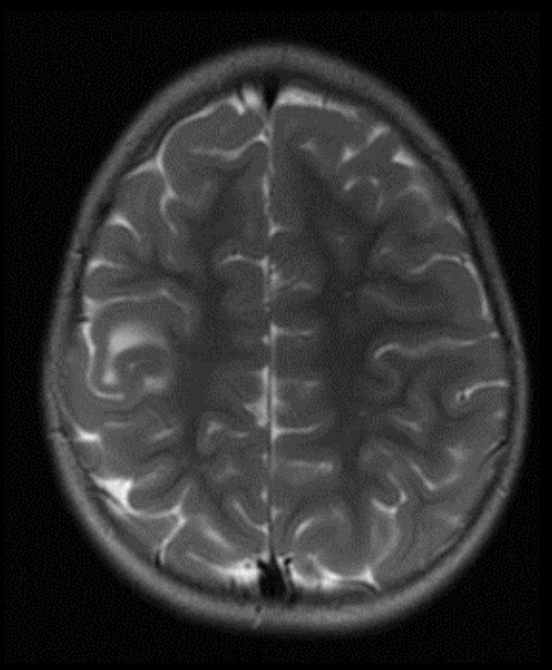




Displasie corticali focali





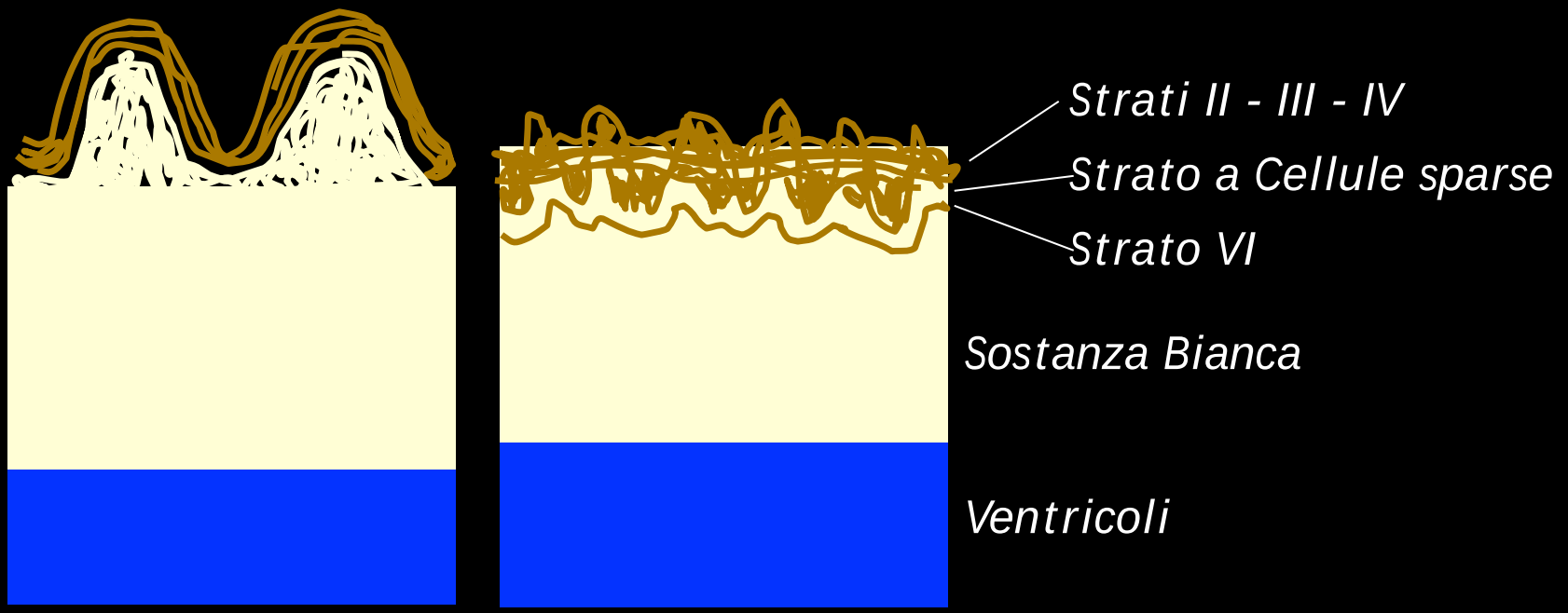


Anomalie della Organizzazione Cellulare

SCHIZENCEFALIE

POLIMICROGIRIE

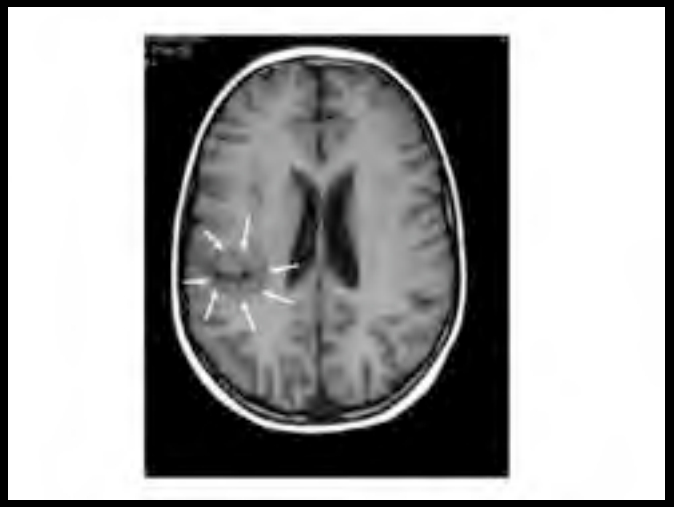
POLIMICROGIRIA

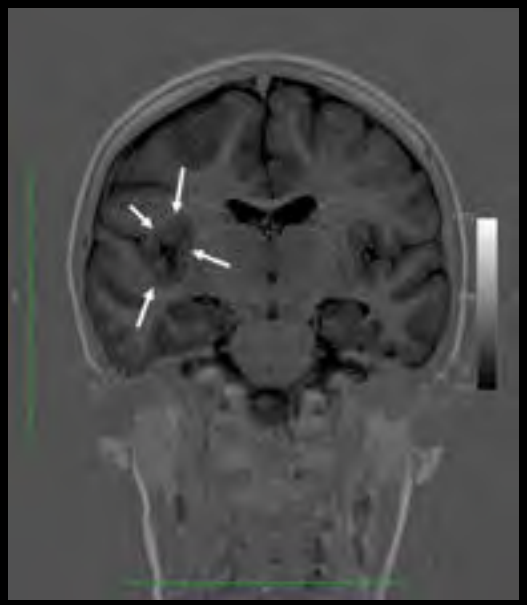


MALFORMAZIONI dello SVILUPPO CORTICALE

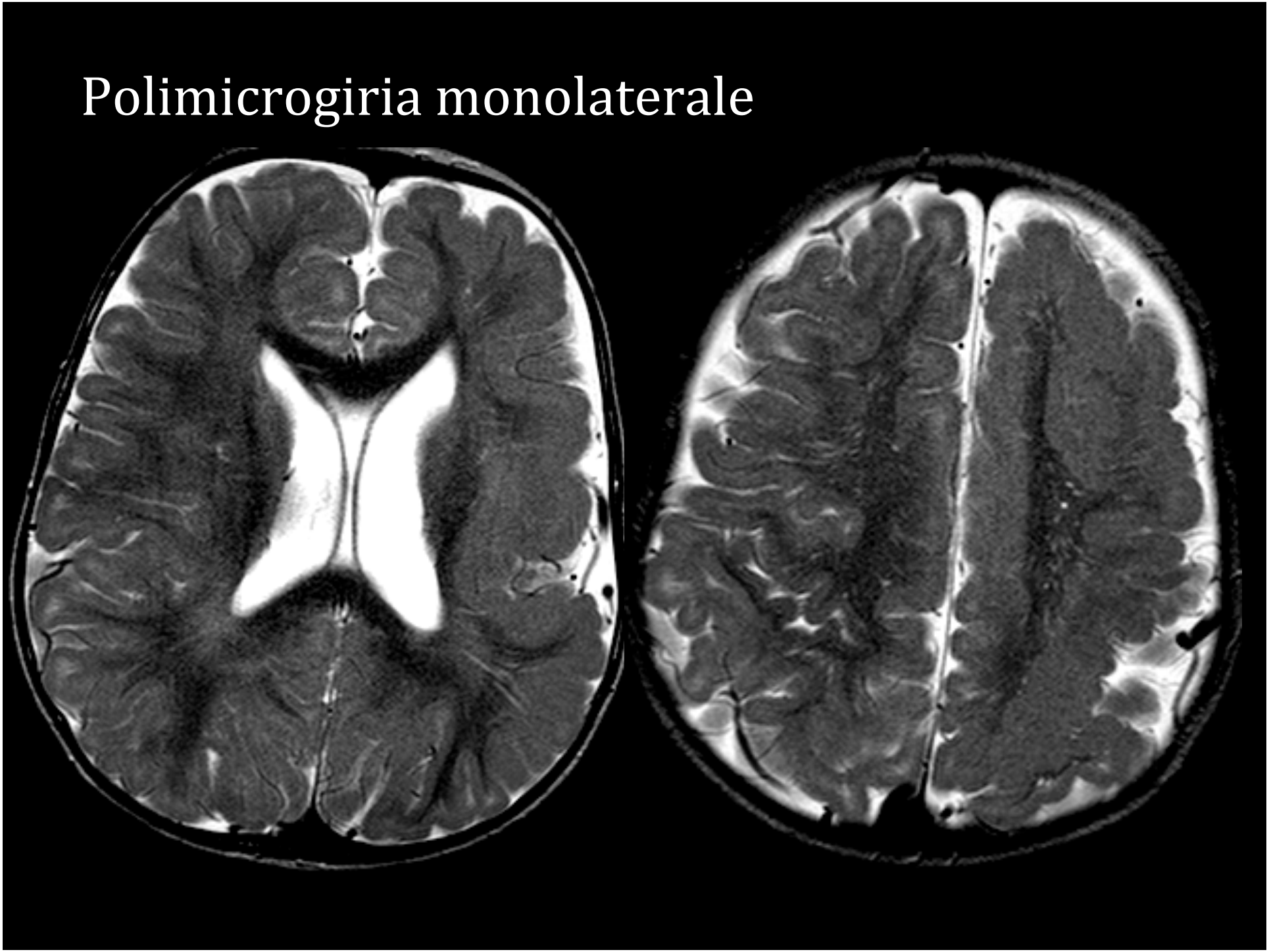
III. MALFORMAZIONI DA ANOMALIE DELL'ORGANIZZAZIONE CORTICALE (inclusa la MIGRAZIONE NEURONALE TARDIVA)

A) POLIMICROGIRIA E SCHIZENCEFALIA





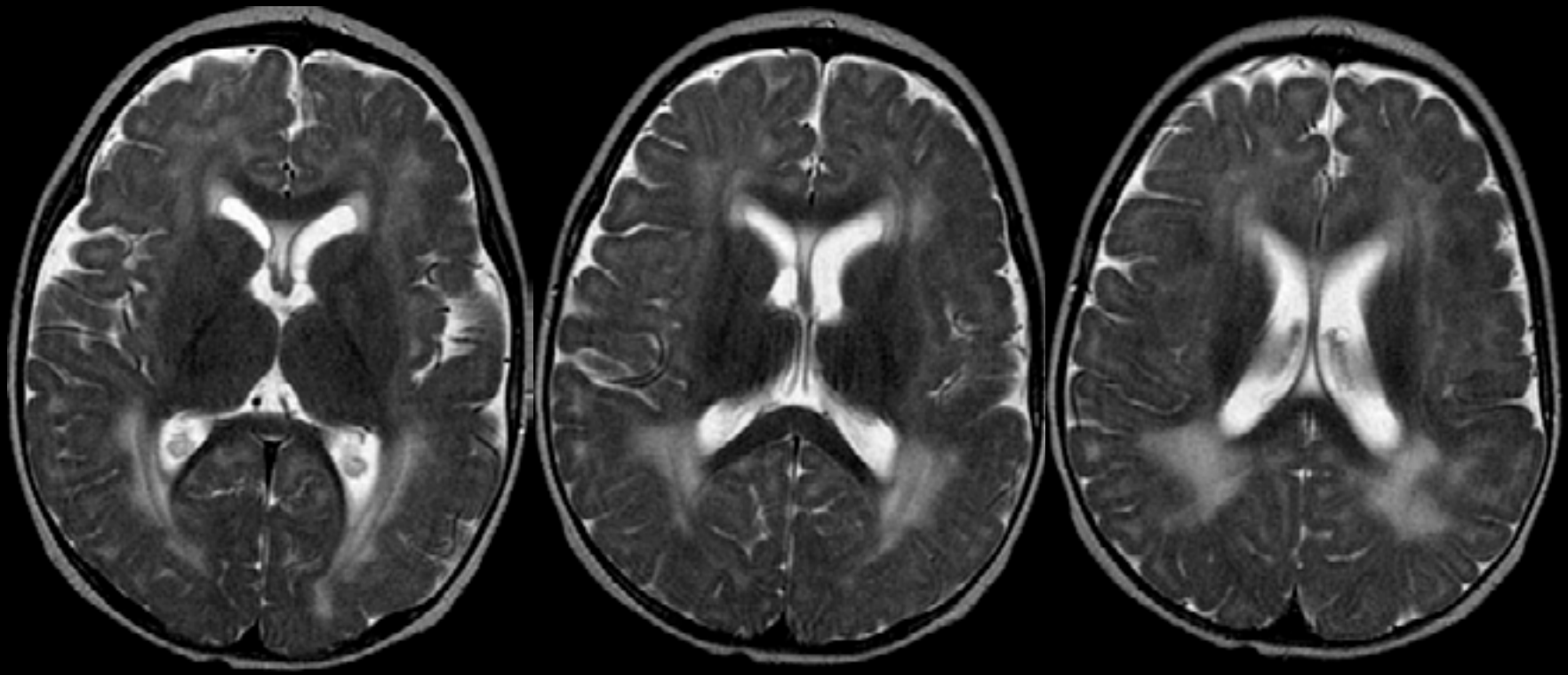
Polimicrogiria monolaterale



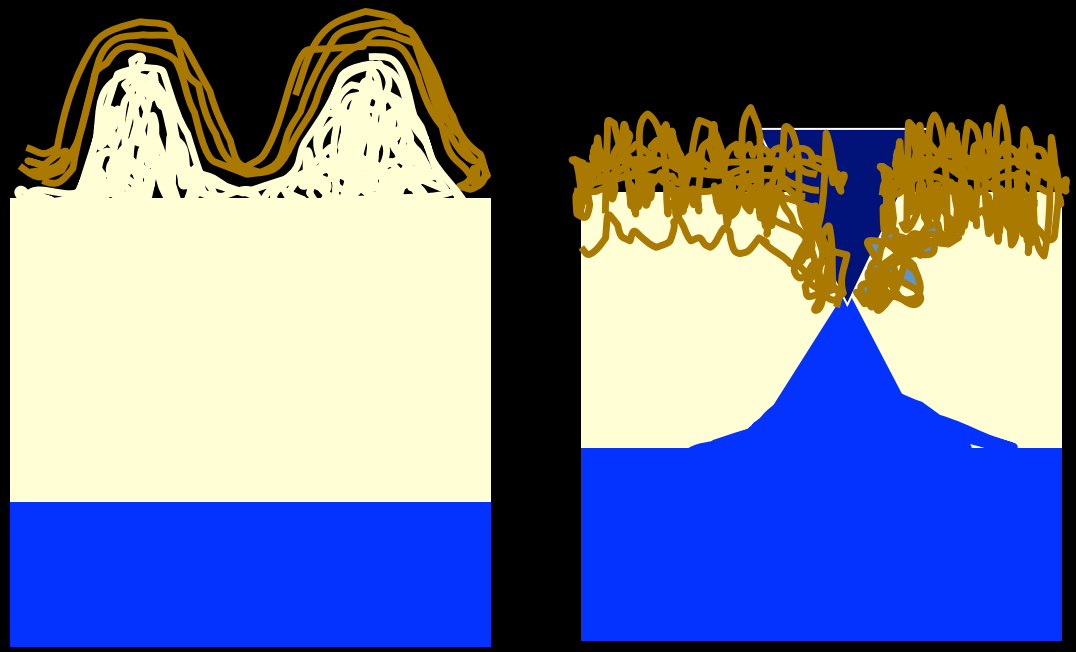
Polimicrogiria bilaterale

Polimicrogiria ACQUISITA

TORCH CMV



SCHIZENCEFALIA



MALFORMAZIONI dello SVILUPPO CORTICALE

III. MALFORMAZIONI DA ANOMALIE DELL'ORGANIZZAZIONE CORTICALE (inclusa la MIGRAZIONE NEURONALE TARDIVA)

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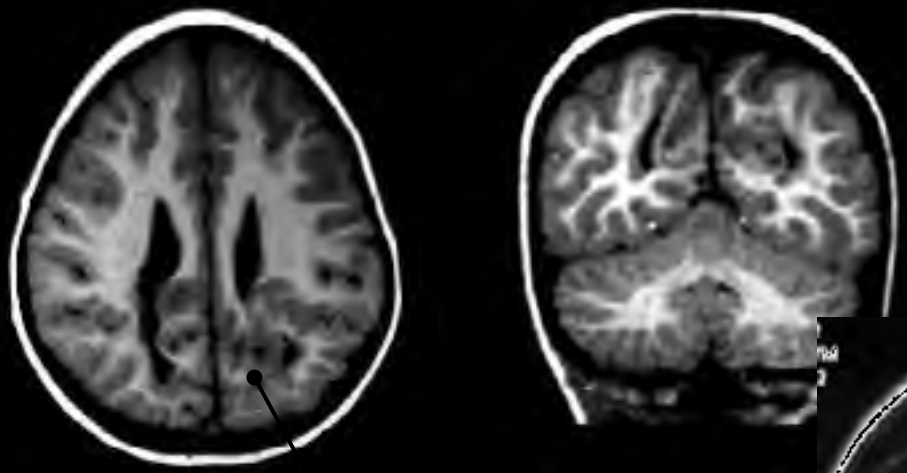
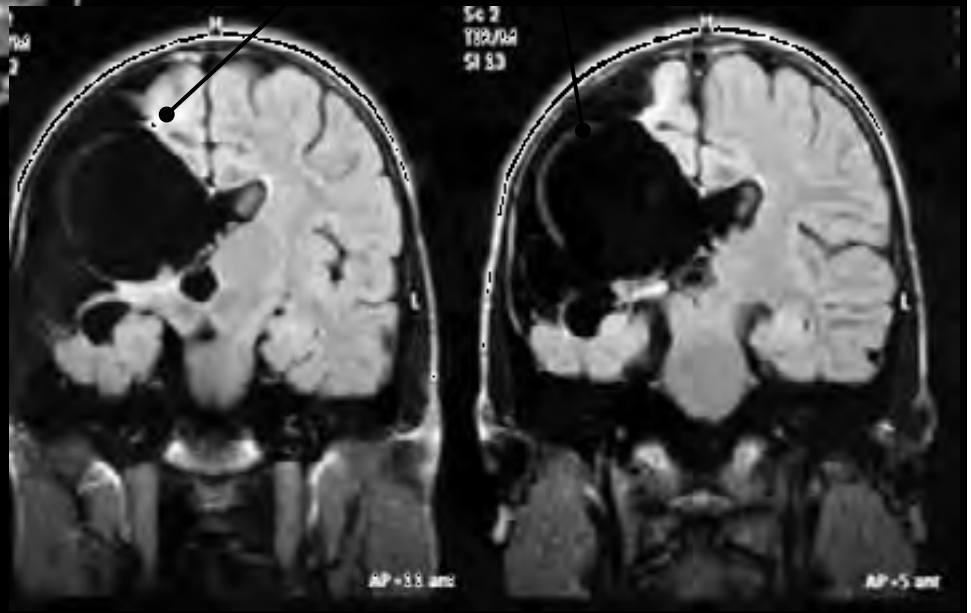


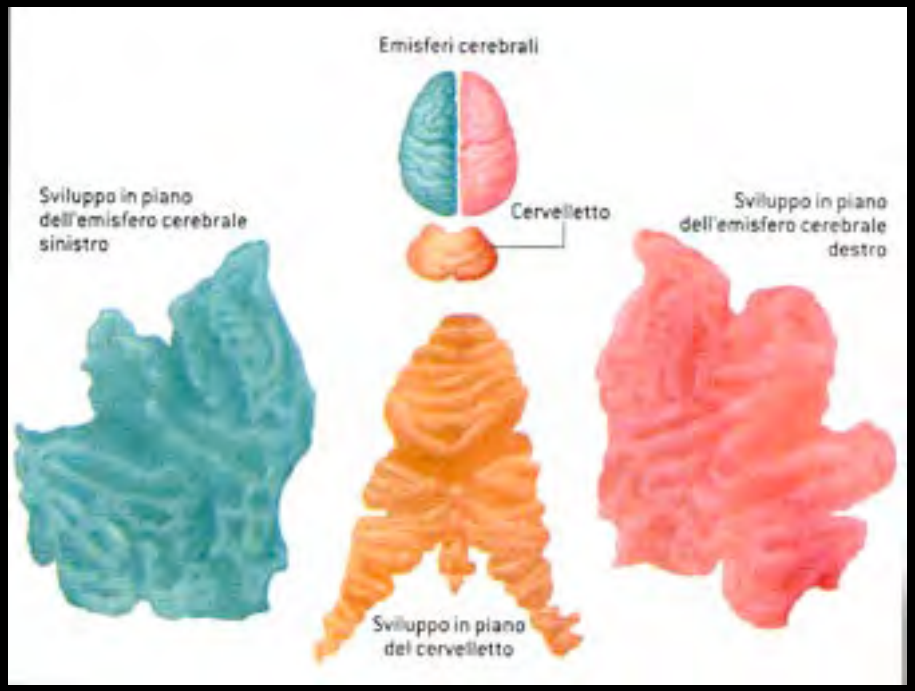
A "labbra chiuse"

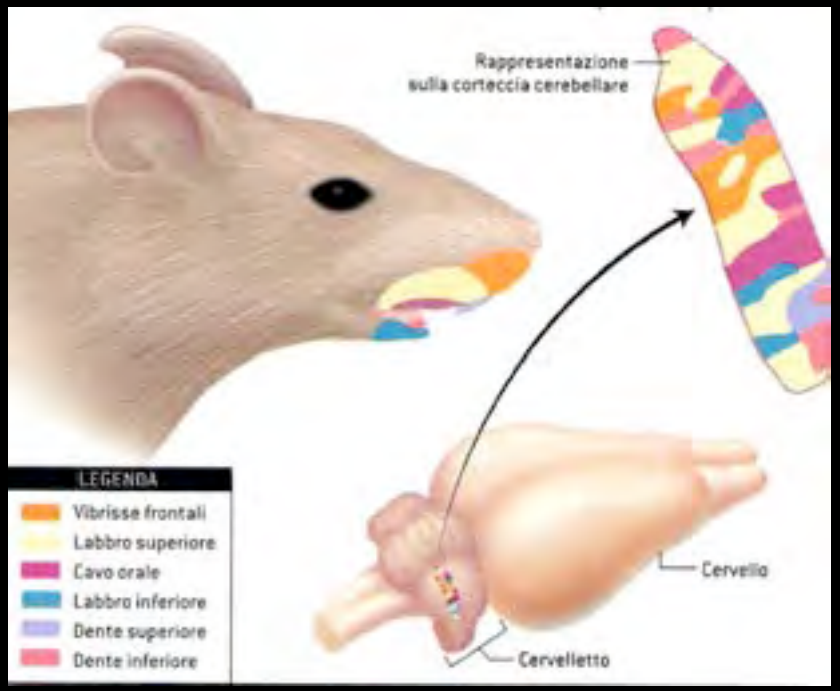
A "labbra aperte"



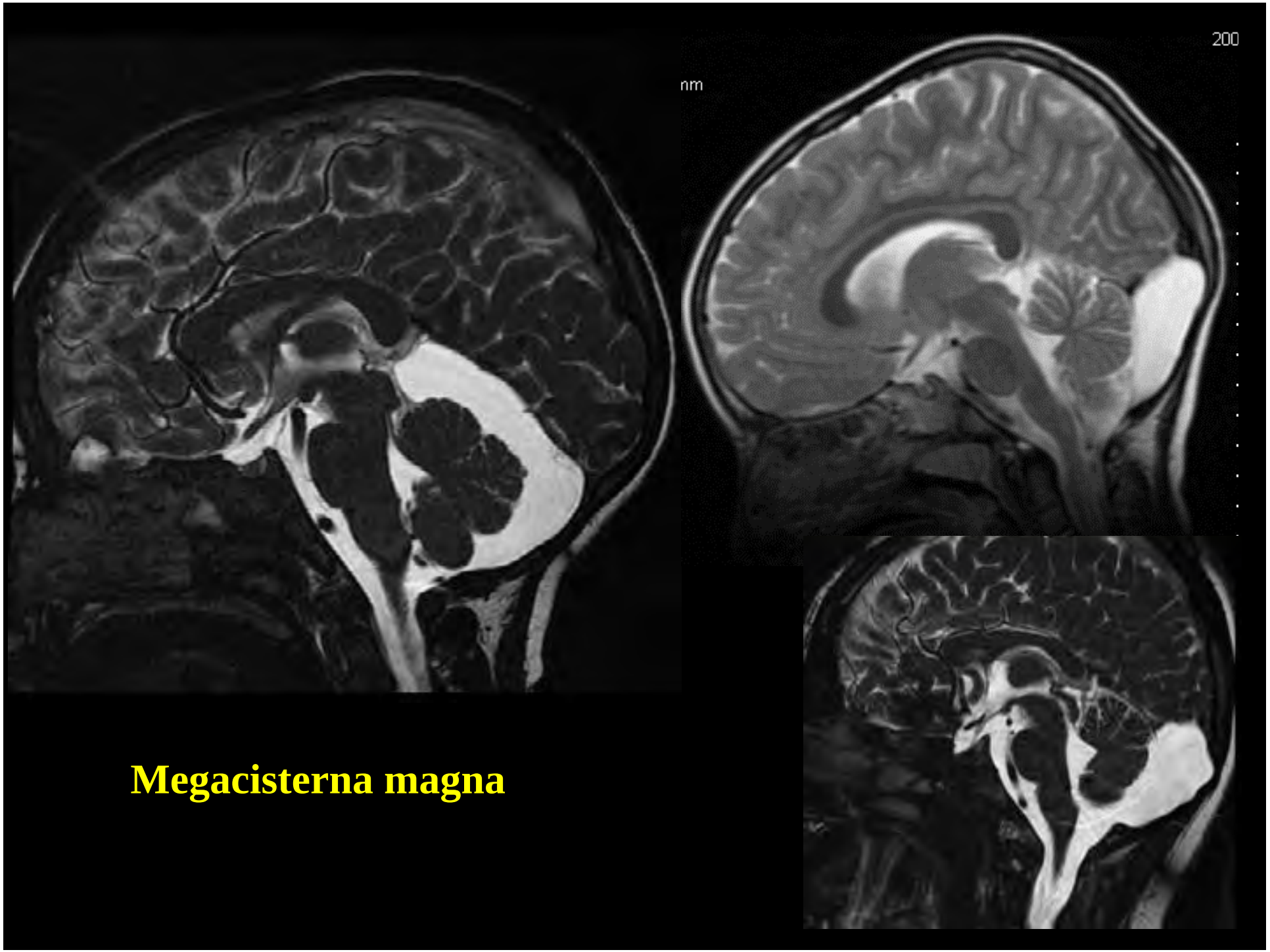

A labbra chiuse
A labbra aperte

Funzione generale e sofisticata di sostegno per il resto del cervello



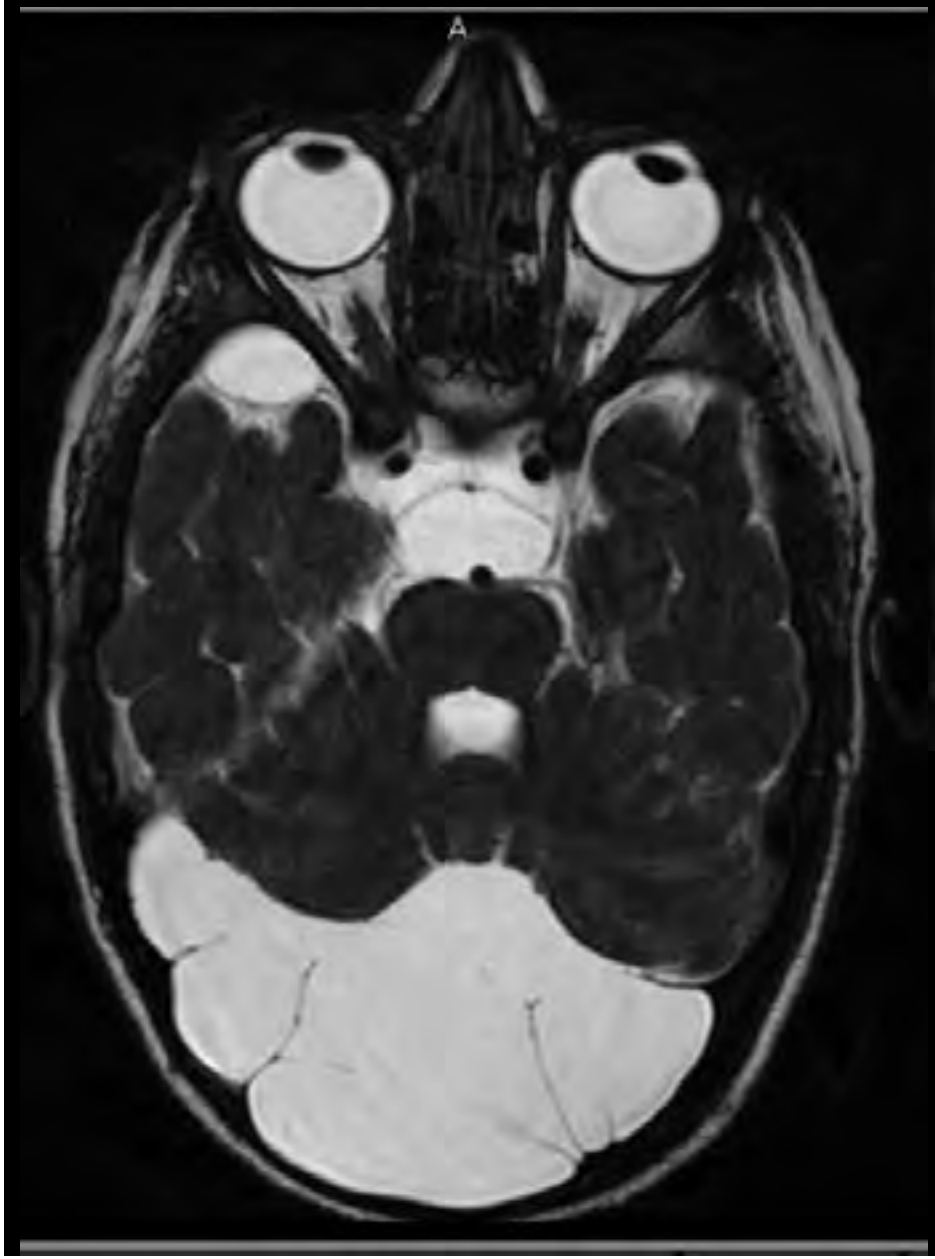


- Controllo / apprendimento movimento ("temporalizzazione generalizzata")
- Controllo della velocità e precisione della percezione di informazioni
- Elaborazione processi di acquisizione segnali sensoriali
- Memoria a breve termine / Memoria di lavoro
- Modulazione delle emozioni / Controllo degli impulsi
- Attenzione / Capacità di apprendimento coordinata
- Funzioni cognitive superiori / Elaborazione informazioni complesse
- Capacità di progettazione rivolta al futuro

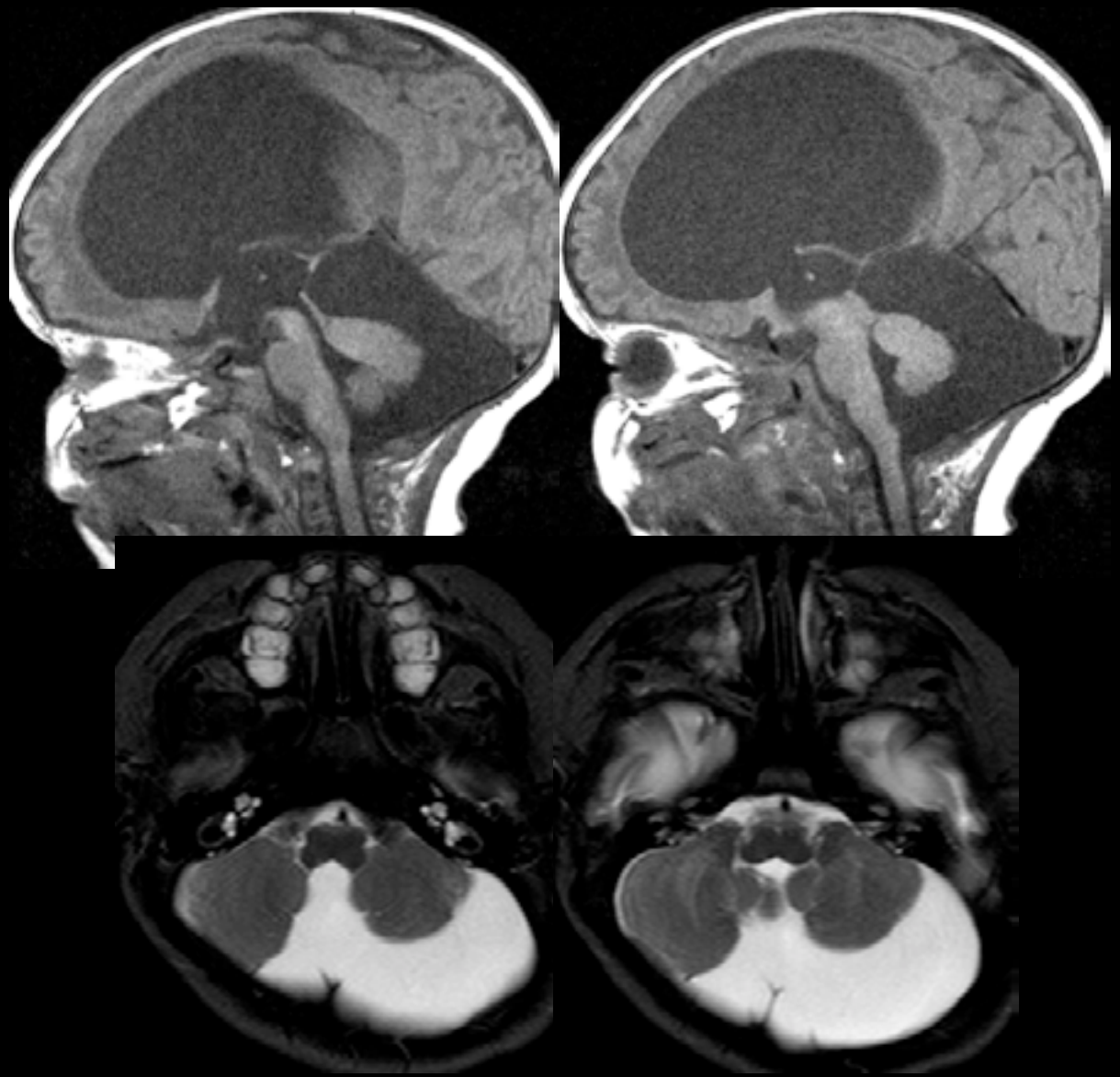


Megacisterna magna

Cisti aracnoidea retrovermiana



Cisti di Blake



MALFORMAZIONI della FOSSA CRANICA POSTERIORE

COMPLESSO DANDY-WALKER (CDW)

Cisti aracnoidea retrocerebellare

Malformazione DW

Ipoplasia VERME CEREBELLARE

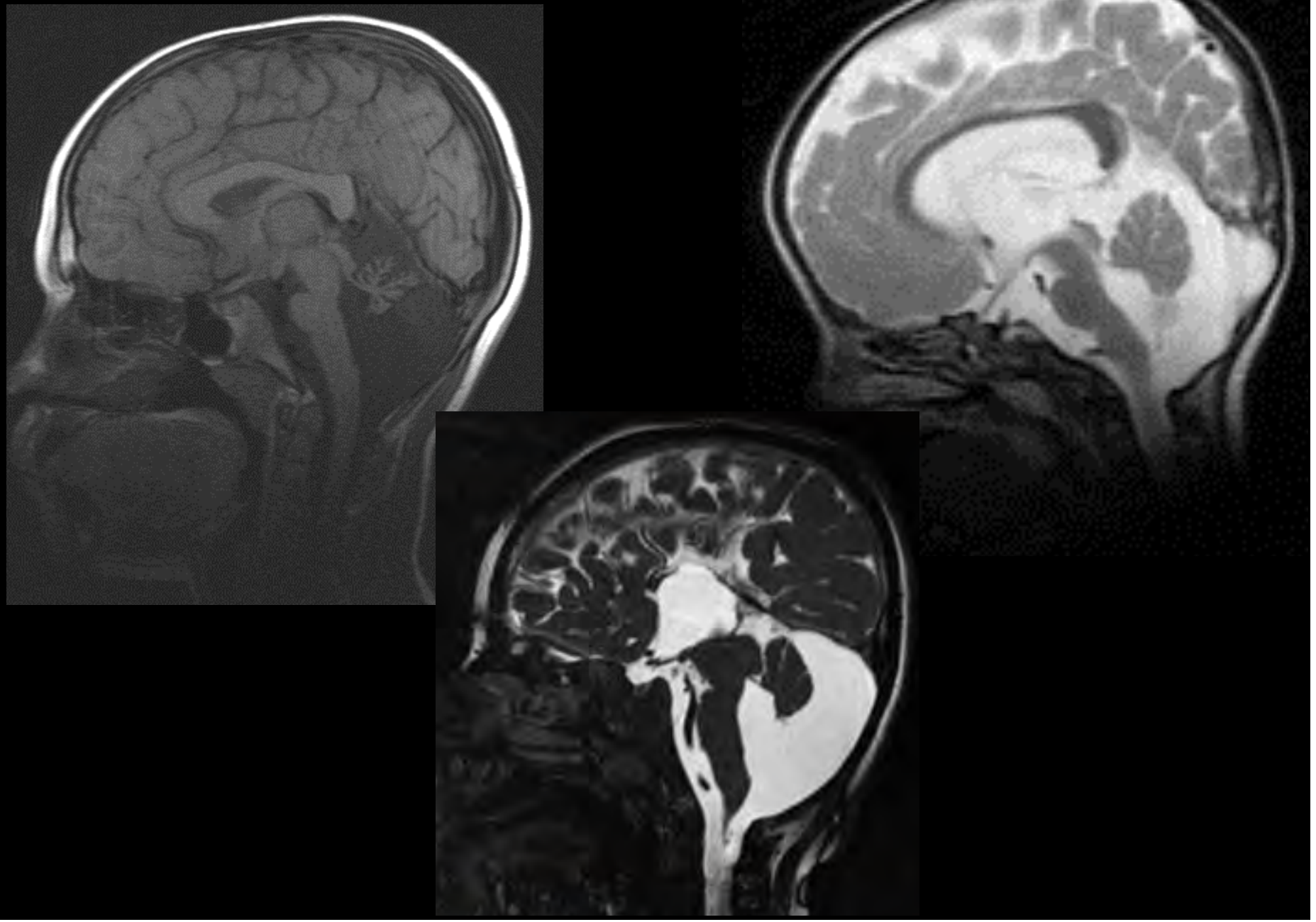
Dilatazione cistica IV VENTRICOLO

Allargamento FOSSA CRANICA POSTERIORE

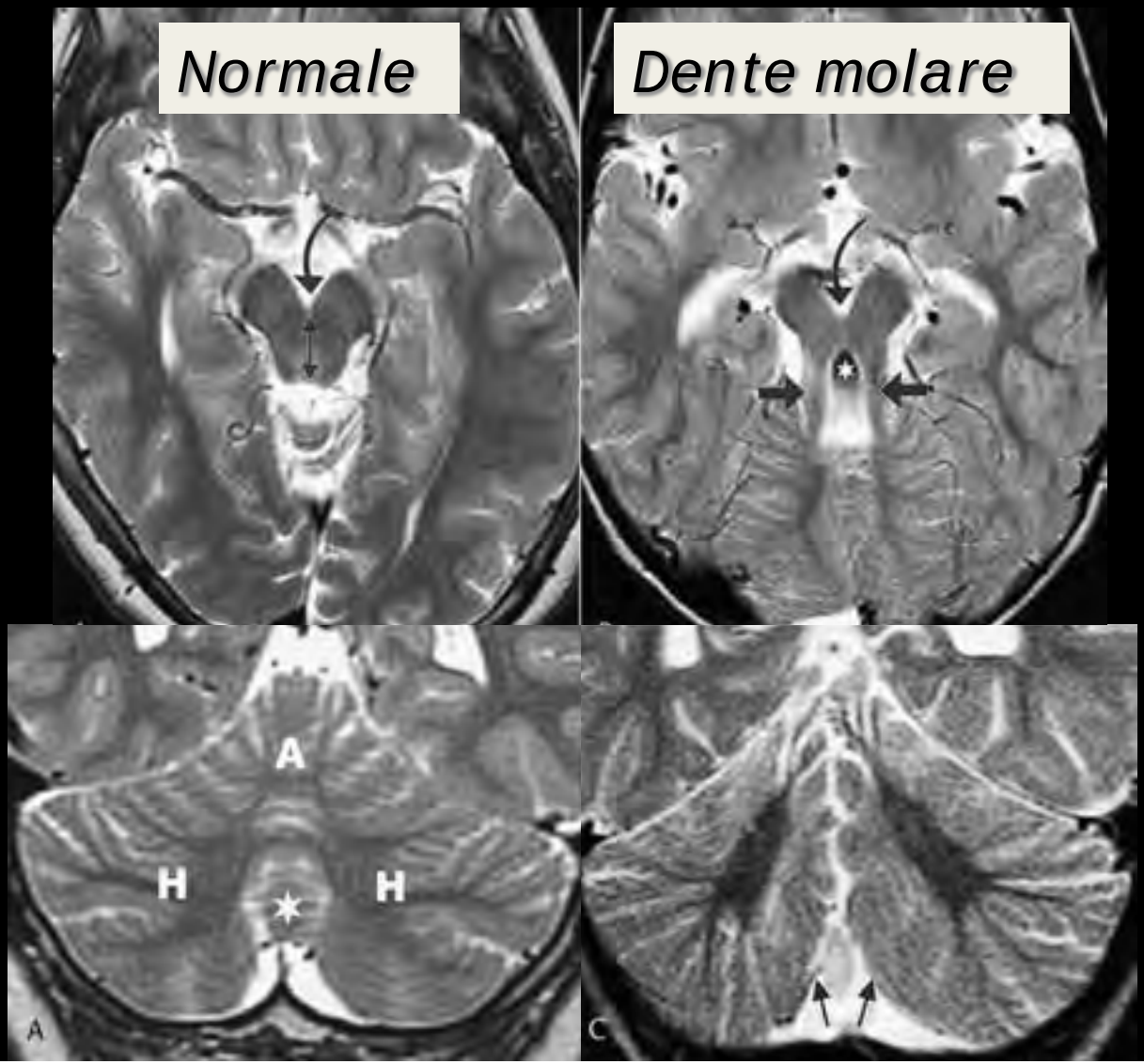
Variante DW

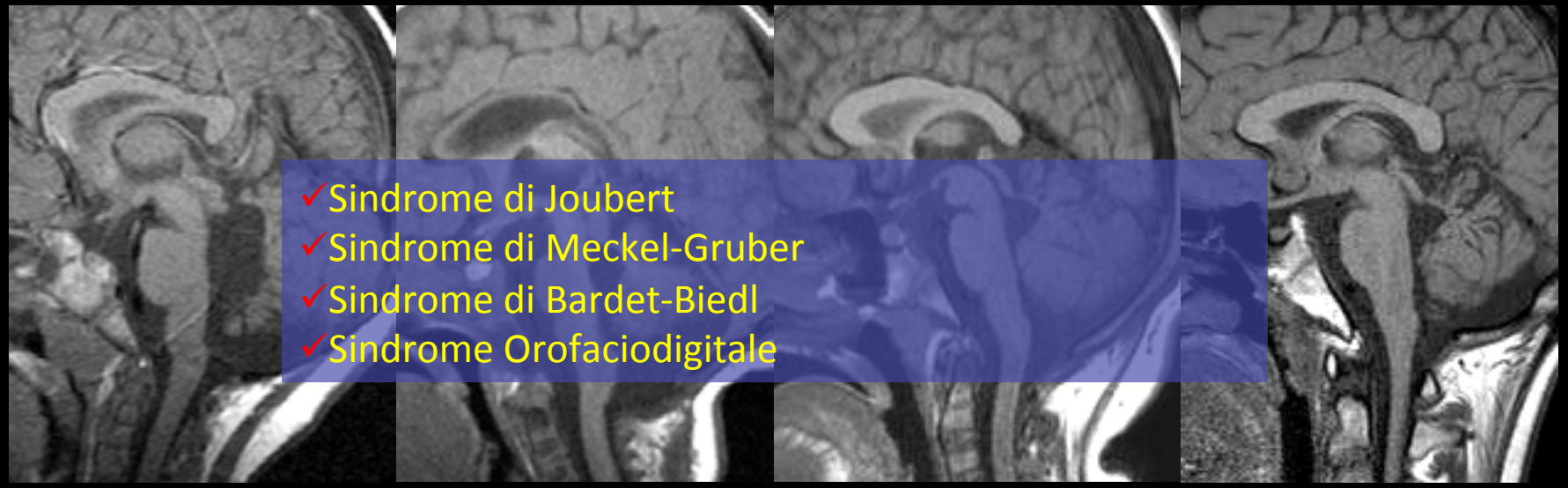
Megacisterna Magna

Malformazione di Dandy-Walker

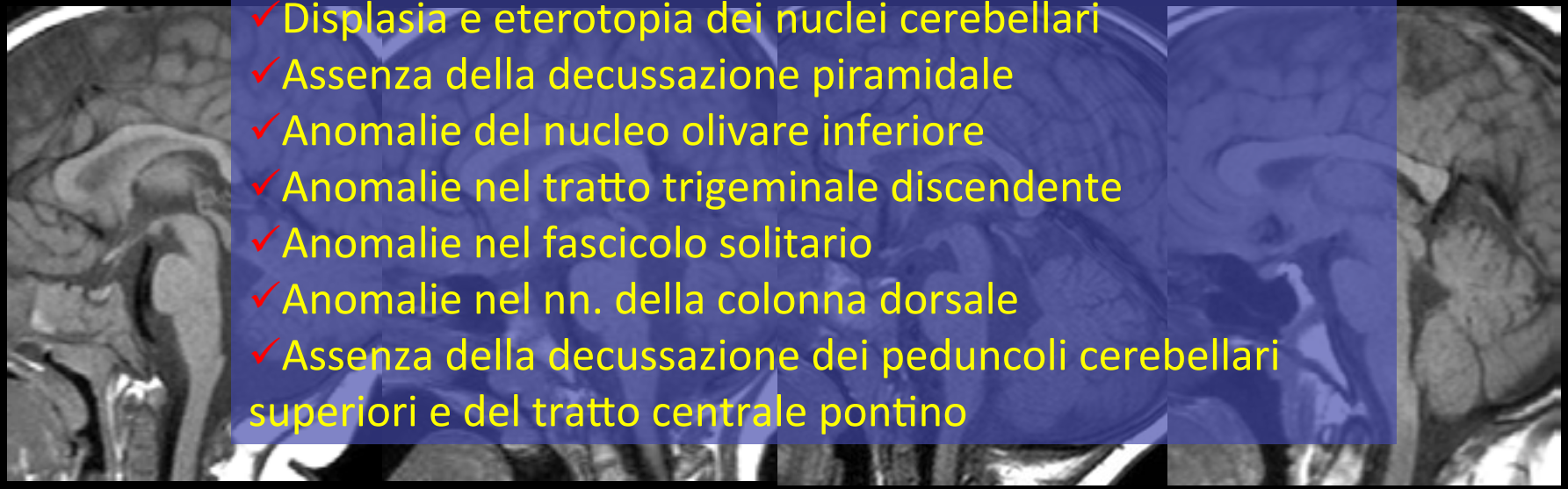


Sindrome di Joubert





- ✓Sindrome di Joubert
- ✓Sindrome di Meckel-Gruber
- ✓Sindrome di Bardet-Biedl
- ✓Sindrome Orofaciodigitale

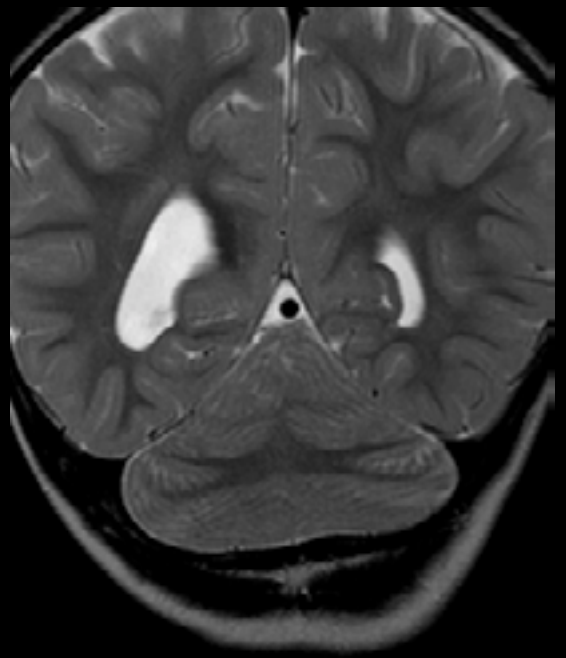


- ✓Displasia e eterotopia dei nuclei cerebellari
- ✓Assenza della decussazione piramidale
- ✓Anomalie del nucleo olivare inferiore
- ✓Anomalie nel tratto trigeminale discendente
- ✓Anomalie nel fascicolo solitario
- ✓Anomalie nel nn. della colonna dorsale
- ✓Assenza della decussazione dei peduncoli cerebellari superiori e del tratto centrale pontino

MALFORMAZIONI CEREBELLARI

ROMBOENCEFALOSINAPSI

Assenza / agenesia VERME CEREBELLARE

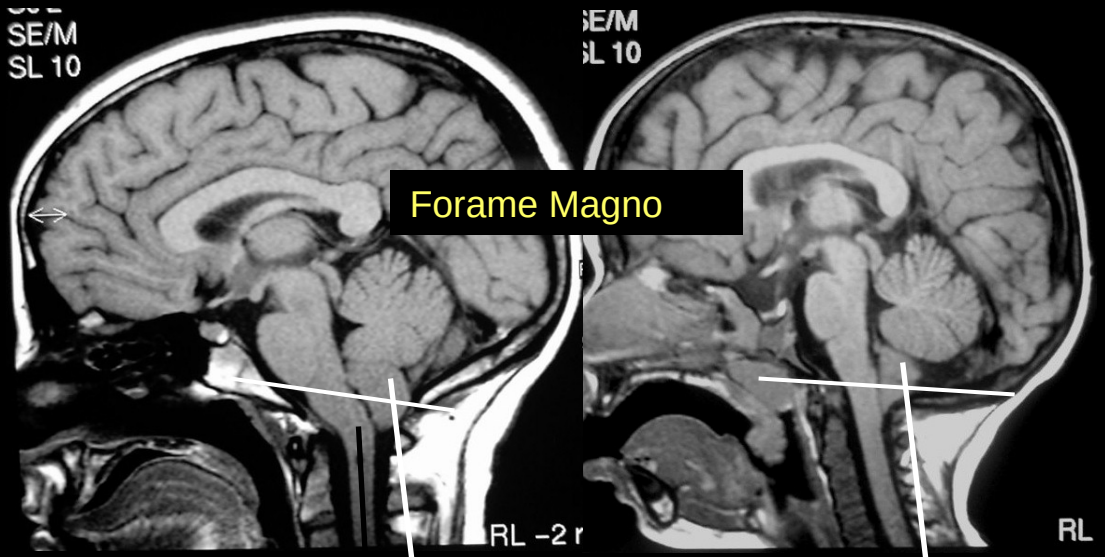


Fusione EMISFERI CEREBELLARI

MALFORMAZIONI della FOSSA CRANICA POSTERIORE

MALFORMAZIONE DI CHIARI

CHIARI 3



CHIARI 1

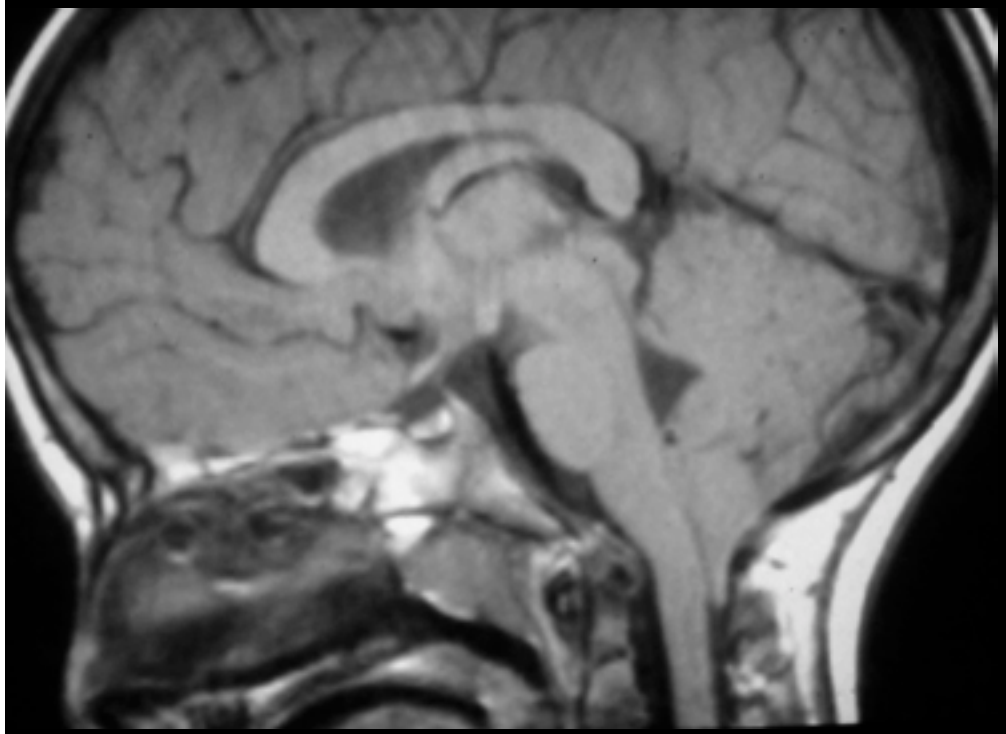
Tonsille cerebellari

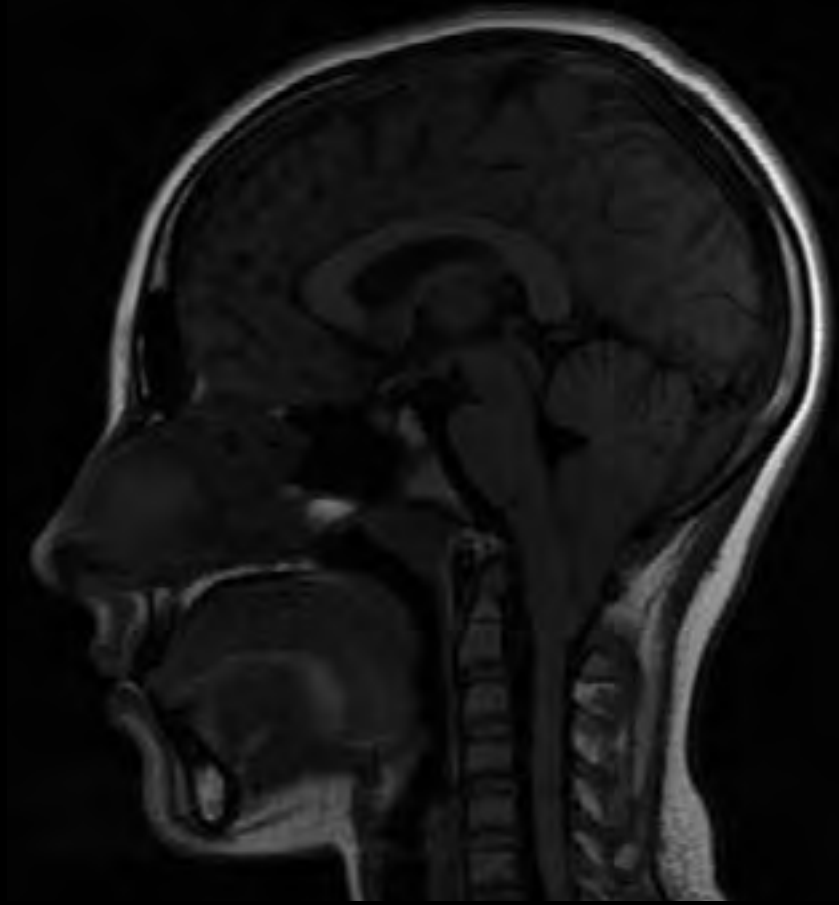
Bulbo e/o ponte

siringomielia

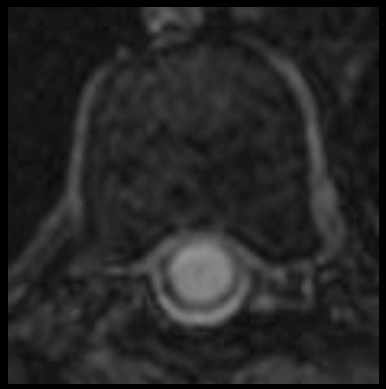
CHIARI 2
Malformazione COMPLESSA

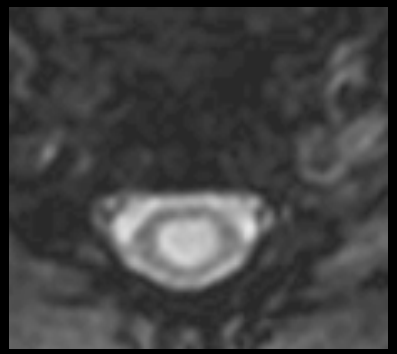
Chiari I





Chiari I siringomielia





Chiari II - mielomeningocele