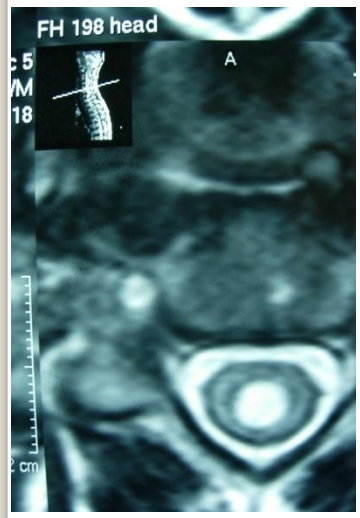
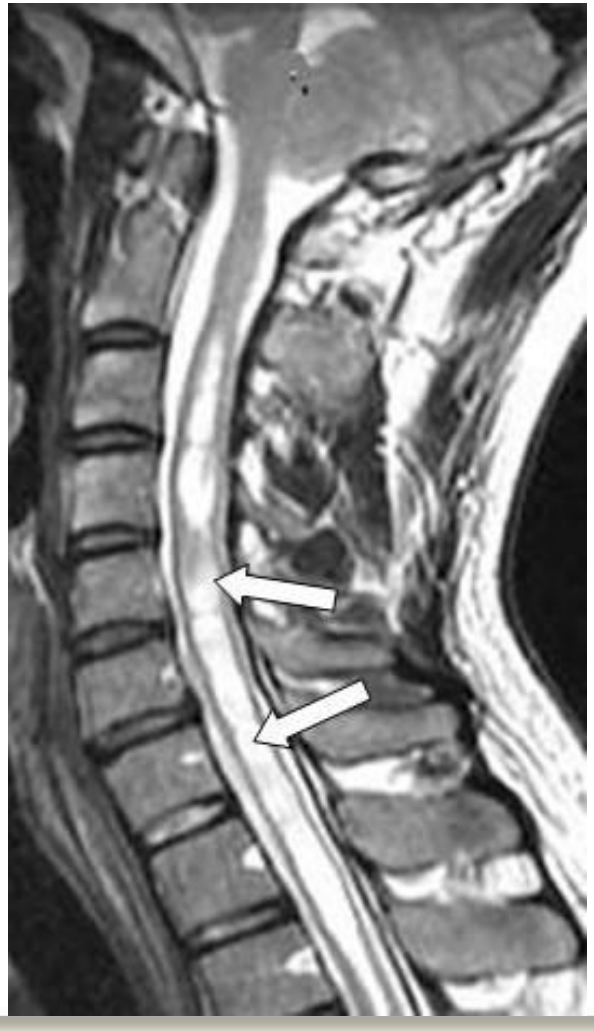
SIRINGOMIELIA E MALFORMAZIONE DI CHIARI 1: SINTOMI E SEGNI CLINICI RILEVANTI

Palma Ciaramitaro
ASLTO5, Torino

Syringomyelia and Syringobulbia

Other diseases of the Spinal Cord
(ORPHACODE 3280; cod. ICD-10 G95.0/Q06.4, cod. ICD-9-CM 336.0)





- PREVALENCE
  2-15:100.000 inhabitants
- AVERAGE AGE
  30-40 years
- SYMPTOMS/SIGNS

- “dissociated" or "suspended“ anesthesia
- "central" neuropathic pain
- flaccid/spastic paresis (II and I Motor Neuron) up to severe tetraparesis
- dysauthonomic impairment
- bulbar signs

Hydromyelia= Other congenital malformations of the spinal cord

REGIONE PIEMONTE

Syringomyelia and Syringobulbia

Classification and Diagnostic Criteria

Type I: with obstruction of the foramen magnum and dilatation of the central spinal canal:

A) Associated to CMI

B) Associated with other obstructive lesions of the foramen magnum

Type II: syringomyelia without obstruction of the foramen magnum, or **idiopathic**.

Type III: syringomyelia with other diseases of the spinal cord:

A) Spinal cord tumours (usually intraspinal)

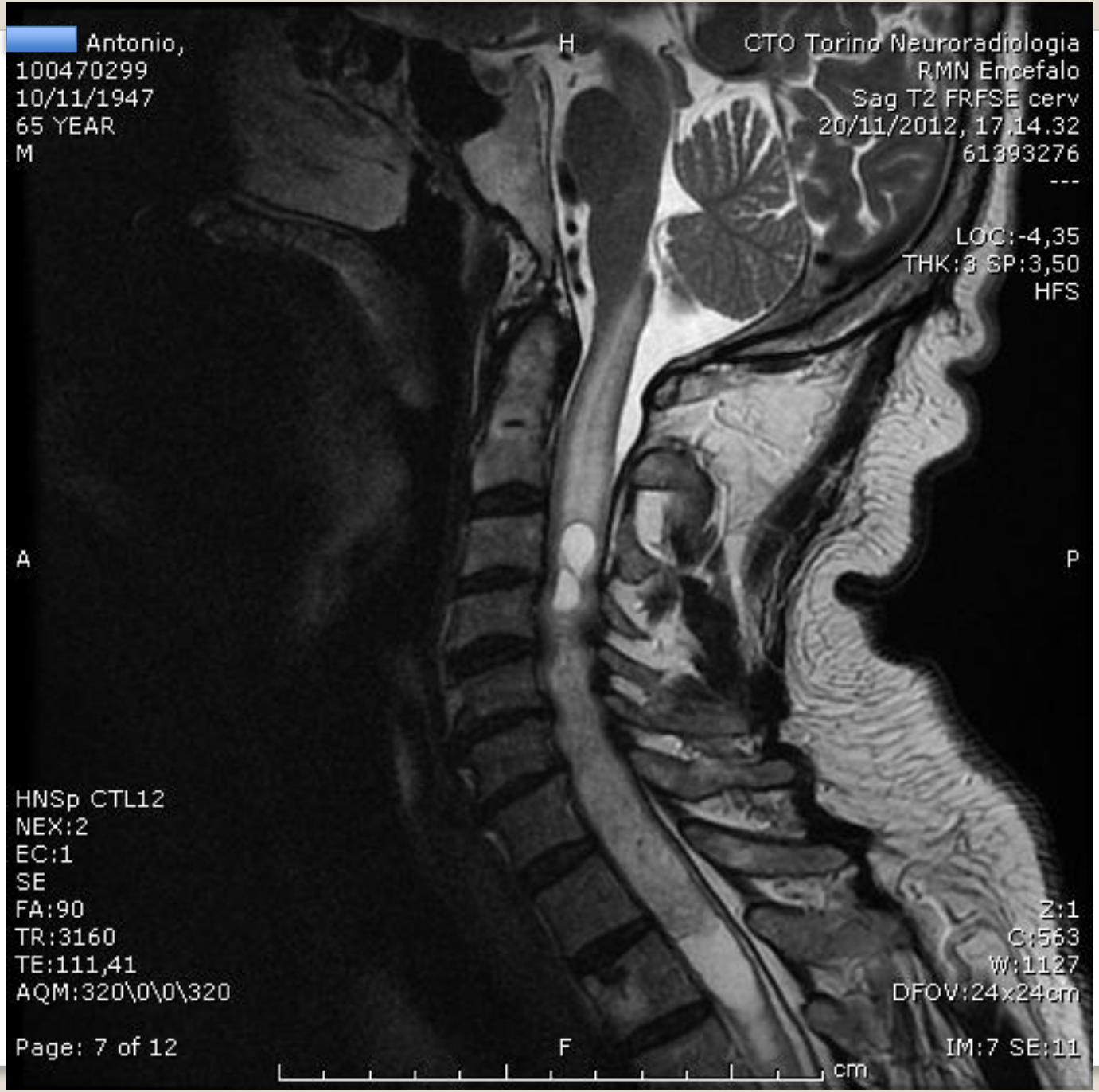
B) Traumatic myelopathy

C) Spinal arachnoiditis and pachymeningitis

D) Myelomalacia due to compression of the spinal cord (tumour, spondylosis).

Type IV: pure hydromyelia, developmental widening of the central canal of the spinal cord, usually associated to hydrocephalus

Diagnosis: syrinx/syringobulbia at MRI, associated with spinal and/or bulbar symptoms and signs related to the cavity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CTO Torino Neuroradiologia
RMN Encefalo
Sag T2 FRFSE cerv
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LOC:-4,35
THK:3 SP:3,50
HFS
A
P
HNSp CTL12
NEX:2
EC:1
SE
FA:90
TR:3160
TE:111,41
AQM:320\0\0\320
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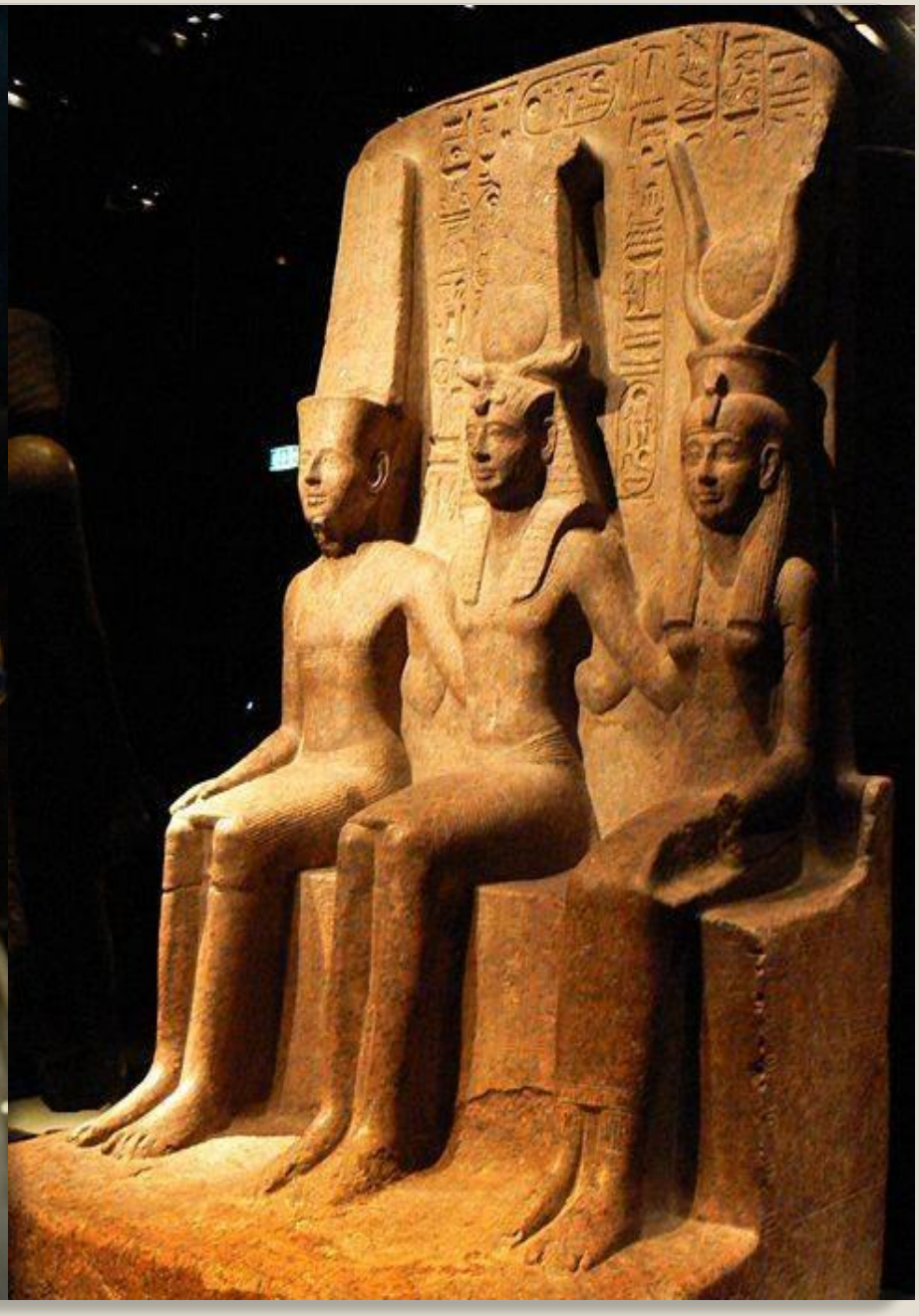
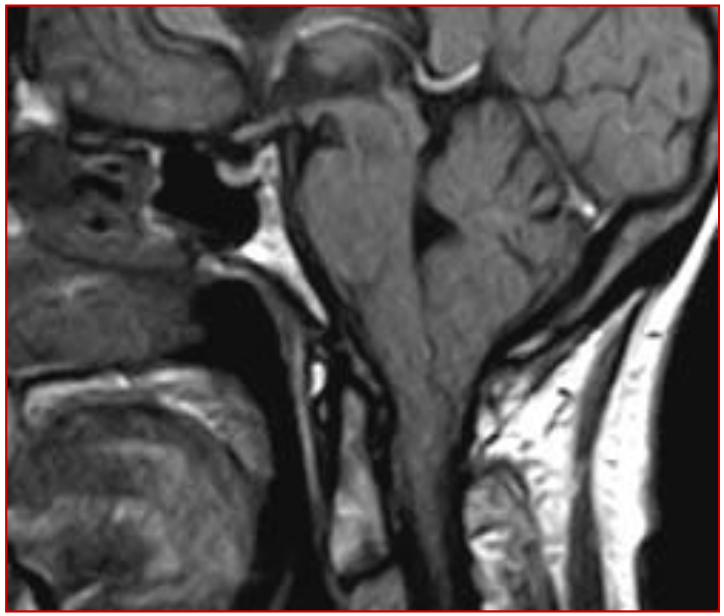
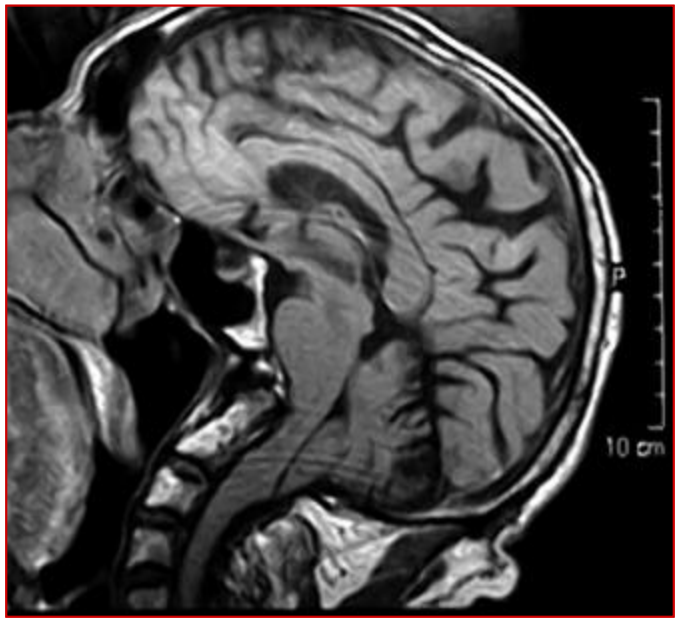


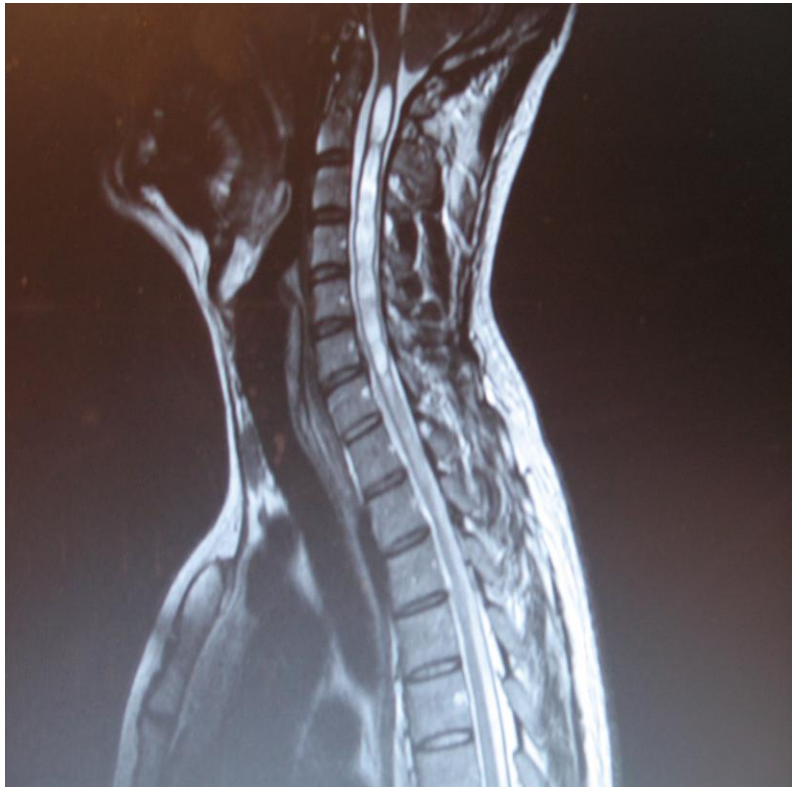
Chiari Malformation

- **Posterior Fossa Malformations**
- **Cerebellar dysplasia: Chiari Malformation**





Chiari Malformations



- **CM I:** paraxial mesoderm disorder, with PCF* abnormalities (mostly small) and the consequent descent of the cerebellar tonsils (children and adults)
  - CMI-A: with Syr at MRI
  - CMI-B: without Syr at MRI
- **CM II:** associated with myelomeningocele (prevalent in children)
- **CM III:** intracranial defects associated with CM II (very rare and severe form)
- **CM IV:** cerebellar aplasia or hypoplasia, associated with tentorium cerebelli aplasia

New!

- **CM O:** CSF flow obstruction (MRI) in absence of cerebellar ectopia; with Syr
- **CM 1.5:** tonsils and brainstem descent

** PCF, posterior cranial fossa*

Arnold-Chiari Syndrome

(ORPHACODE 268882; cod. ICD-10 Q07; ICD-9-CM: Type I 348.4)

Diagnostic Criteria

Chiari I Malformation (neuroradiological) definition

According to **IHS diagnostic criteria** (the second updated edition of "International Classification of Headache Disorders", code 7.7)

Cerebellar tonsillar herniation is defined by one of the following on craniocervical MRI:

- ≥ 5 mm caudal descent of the cerebellar tonsils
- ≥ 3 mm caudal descent of the cerebellar tonsils plus at least one of the following indicators of crowding of the subarachnoid space in the area of the craniocervical junction:
  - -compression of the CSF spaces posterior and later to the cerebellum
  - -reduced height of the supraocciput
  - -increased slope of the tentorium
  - -kinking of the medulla oblongata

Health Council Circular 23/11/2011

REGIONE PIEMONTE

Chiari Syndrome

Definition and Clinical Criteria

Chiari Syndrome is the clinical manifestation (symptoms and signs) of Chiari Malformation (radiologically defined)= "symptomatic Chiari".

❖**Headache**, according to IHS diagnostic criteria characterised by at least one of the following and fulfilling criteria:

-precipitated by cough and/or Valsalva manoeuvre

-protracted (hours to days) occipital and/or sub-occipital headache

❖**Symptoms and/or signs of brainstem, cerebellar and/or cervical cord dysfunction**

❖**Otoneurogical symptoms and/or signs** (eg, dizziness, disequilibrium, sensations of alteration in ear pressure, hypacusia or hyperacusia, vertigo, down-beat nystagmus, oscillopsia).

❖**Transient visual symptoms** (spark photopsias, visual blurring, diplopia or transient visual field deficits).

❖Demonstration of **clinical signs** relevant to **cervical cord**, **brainstem or lower cranial nerves** or of **ataxia** or **dysmetria**.

Health Council Circular 23/11/2011

REGIONE PIEMONTE

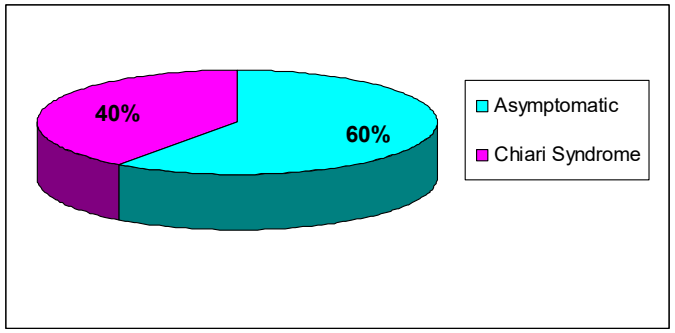
What is really caused by Chiari?

Clinical diagnosis – part I: What is really caused by Chiari I

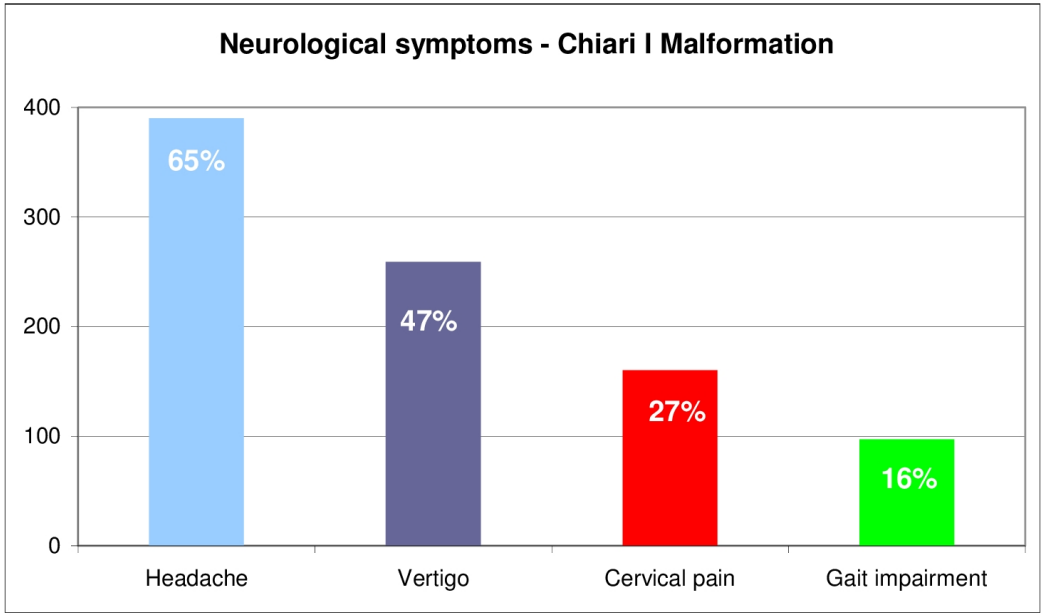
Ciaramitaro Palma, Ferraris Marilena, Massaro Fulvio, Garbossa Diego

Child's Nervous System. 2019 doi.org/10.1007/s00381-019-04206

CHIARI I MALFORMATION (CRESSC series)



N=600
females=73% (N=440)
Age>18 yrs= 98%
CM1-B (+Syr)= 56% (25% asymp)
CM1-A= 44% (60% asympt)



Clinical diagnosis – part I: What is really caused by Chiari I

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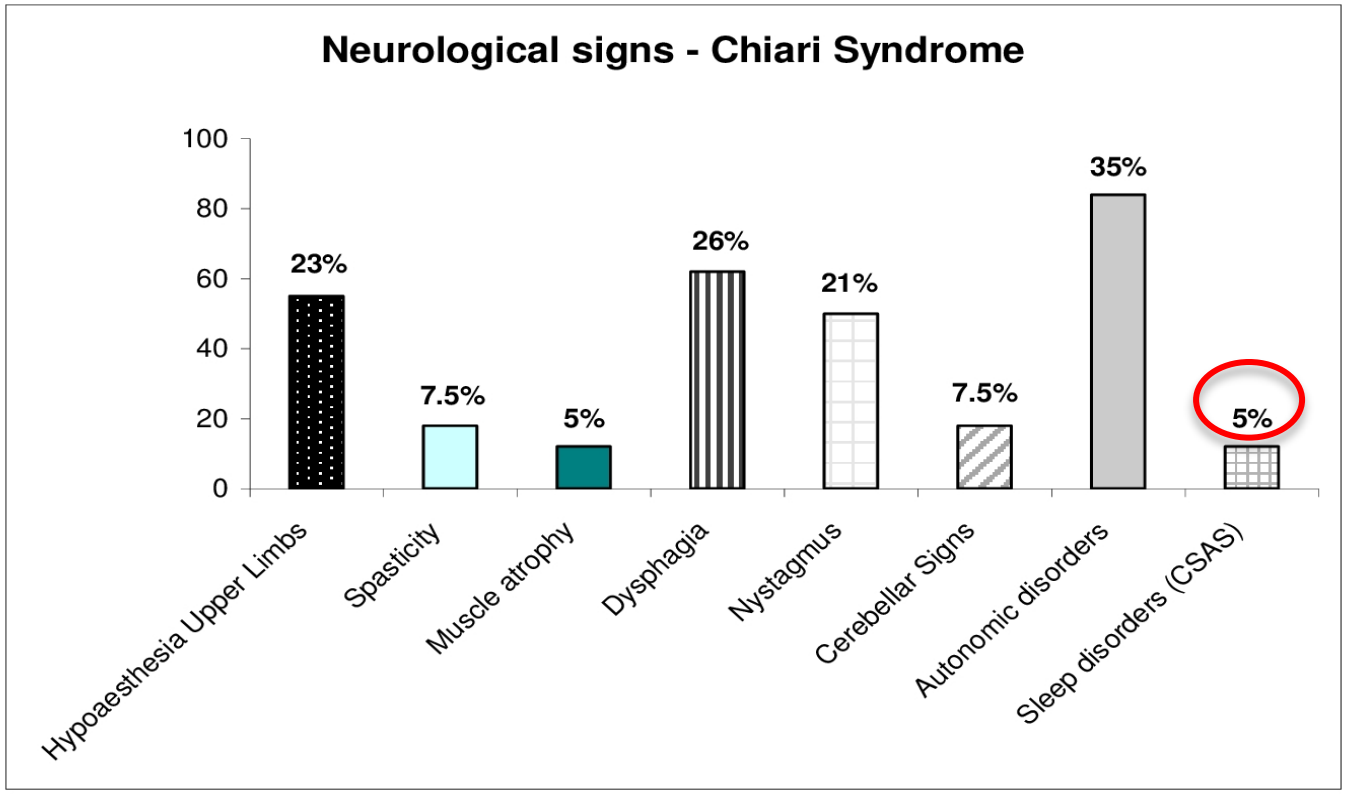

Ann Ist Super Sanità 2020 | Vol. 56, No. 1: 48-58
DOI: 10.4415/ANN_20_01_08

Syringomyelia and Chiari Syndrome Registry: advances in epidemiology, clinical phenotypes and natural history based on a North Western Italy cohort

Palma Ciaramitaro[1], Diego Garbossa[2], Paola Peretta[3], Gianluca Piatelli[4], Luca Massimi[5], Laura Valentini[6], Giuseppe Migliaretti[7], Simone Baldovino[8], Dario Roccatello[8], Yllka Kodra[9], Domenica Taruscio[9], on behalf of the Interregional Chiari and Syringomyelia Consortium*

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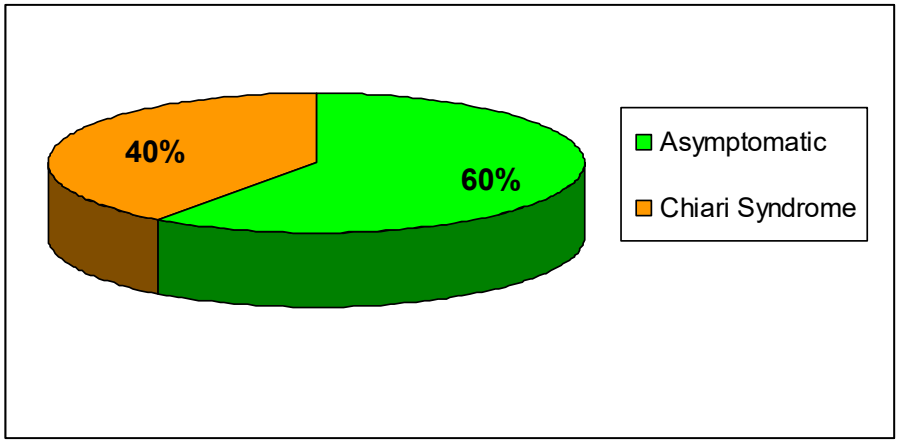
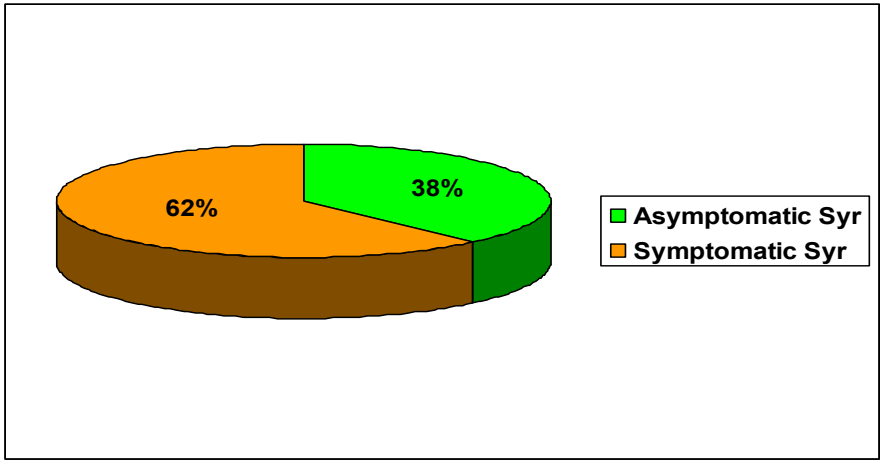
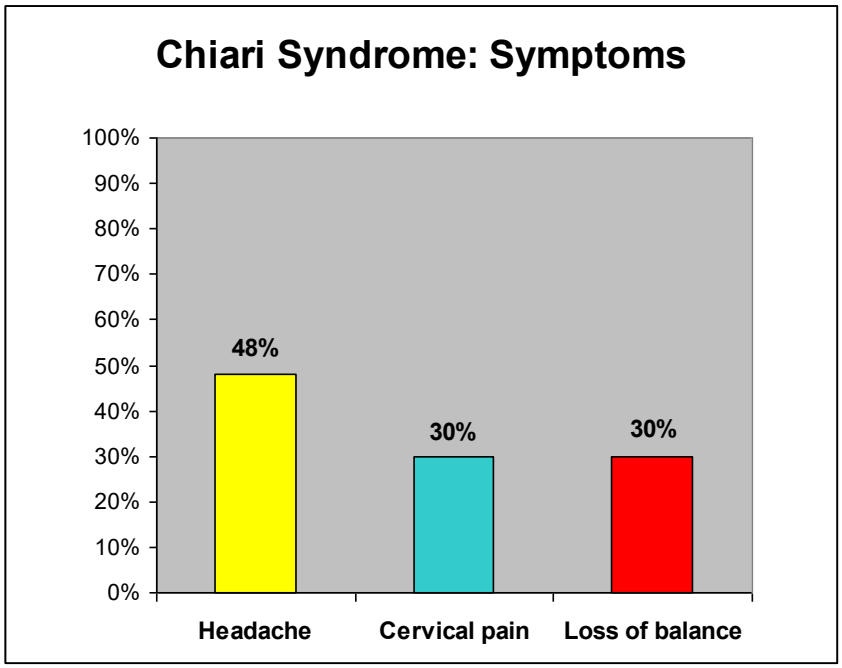
[9]Centro Nazionale Malattie Rare, Istituto Superiore di Sanità, Rome, Italy
**the members of the Interregional Chiari and Syringomyelia Consortium are listed before the references*

CHIARI MALFORMATION

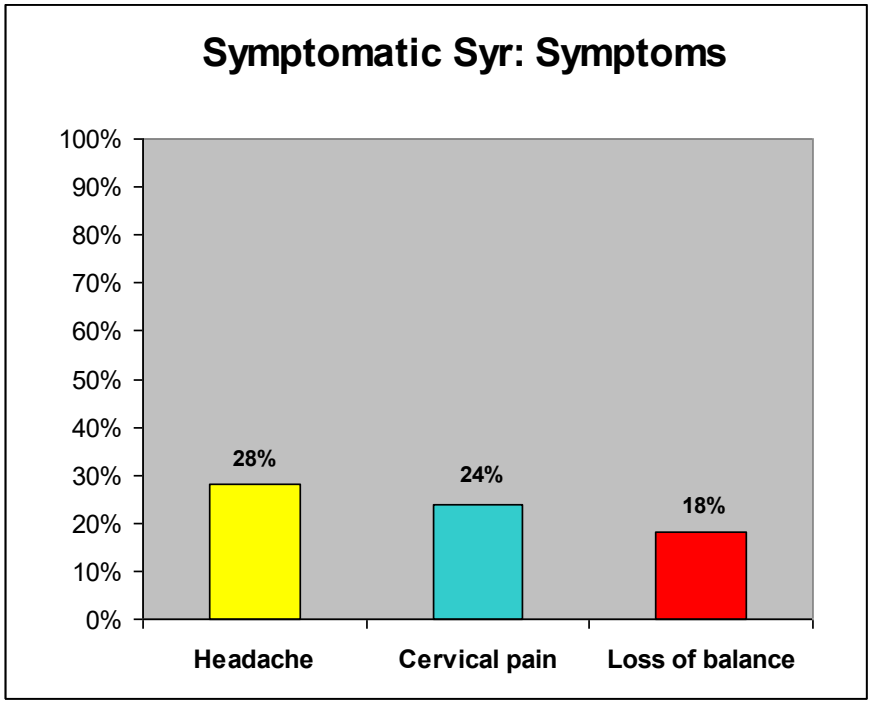


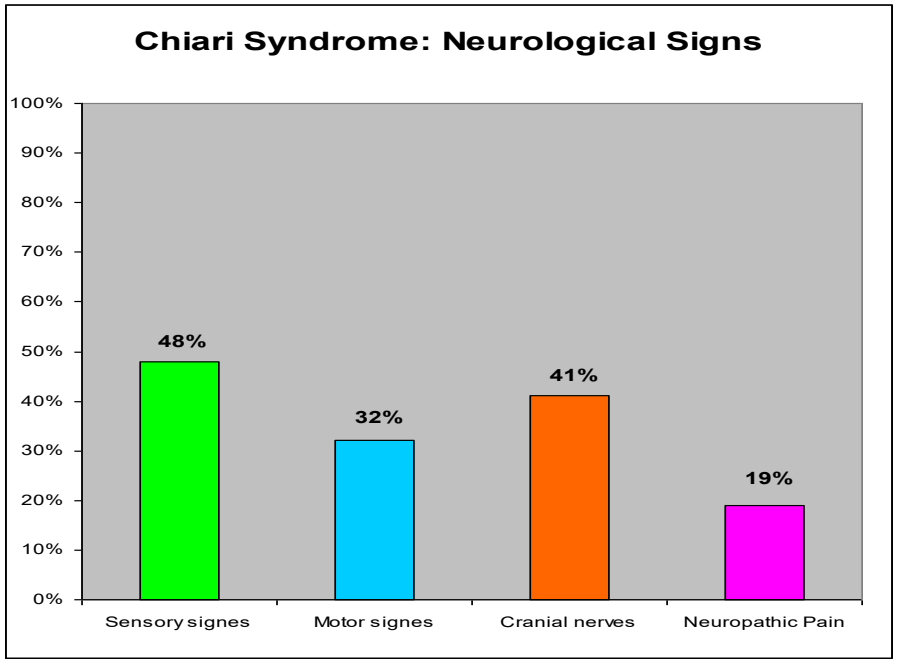
SYRINGOMYELIA

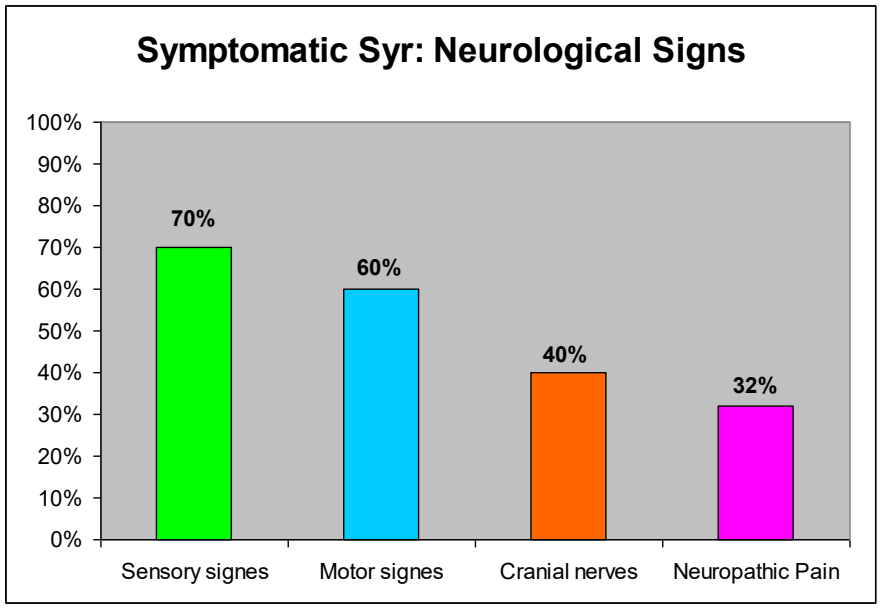



Chiari Syndrome: Symptoms
100%
90%
80%
70%
60%
50%
40%
30%
20%
10%
0%
48%
30%
30%
Headache
Cervical pain
Loss of balance

SINTOMI


Symptomatic Syr: Symptoms
100%
90%
80%
70%
60%
50%
40%
30%
20%
10%
0%
28%
24%
18%
Headache
Cervical pain
Loss of balance





SEGNI

Hypoaesthesia UL/LL
Paraesthesia
Sensory Levels

Hyposthenia UL/LL
Upper /Lower MTN signs
Spasticity
Paraparesis
Tetraparesis
Paraplegia
Tetraplegia

Dysphagia
Nystagmus

CM patients

	Total (N=347)	Chiari Syndrome (40% N=139)	CM Asymptomatic (60% N=208)
Age at diagnosis (mean±SD)	34±19,7	36±20,6	33±19,1
Age at survey (mean±SD)	37±20,9	40±21,5	36±19,7
Sex (%)			
Male	32	47	53
Female	68	37	63
Morphology (%)			
Tonsillar herniation ≥5mm	93	42	58
Tonsillar herniation 3-4mm	4	17	83
Tonsillar herniation ≤3 mm	3	0	100
Types (%)			
CMI A - CMI+Syr	36	60	40
CMI B-isolated	58	25	75
CMII + Myelomeningocele	1	100	0
Other Associated Conditions*	5	59	41
Estimated Prevalence/100.000 (CI 95% range)	**7,5** (6,792-8,407)	**3,0** (2,55-3,58)	**4,5** (3,94-5,19)
Estimated Incidence/100.000 (CI 95% range)**	**3** (2,528-3,556)	**1,2** (0,904-1,560)	**1,8** (1,44-2,24)

*Other associated conditions: 41% Retroflexed Odontoid, 24% Hydrocephalus, 24% Klippel-Feil , 11% Tethered Cord Syndrome

**new reported cases in 2011

Syr patients

	Total (N=217)	Symptomatic (62% N=135)	Asymptomatic (38% N=82)
Age at diagnosis (mean±SD)	39±18,06	38±17,79	40±18,56
Age at survey (mean±SD)	45±18,96	46±18,81	44±19,31
Sex (%)			
Male	36	64	36
Female	64	61	39
Morphology (%)			
Syr	73	78	22
Hydro	27	20	80
Distribution (%)			
Syringobulbia	1	100	0
Syr /Hydro cervical	25	61	39
Syr /Hydro thoracic	24	23	77
Syr /Hydro cervical-thoracic	50	81	19
Etiology (%)			
Type I-primary Syr	59	74	26
Type II-idiopathic Syr	9	50	50
Type III-secondary Syr	14	77	23
Type IV - pure Hydro	18	18	82
Estimated Prevalence /100.000 (CI 95% range)	**4,7** (4,124- 5,406)	**2,9** (2,47-3,49)	**1,8** (1,422-2,220)
Estimated Incidence */100.000 (CI 95% range)	**0,8** (0,568-1,112)	**0,5** (0,318-0,753)	**0,3** (0,167-0,512)

***new reported cases in 2011**

Neurological Sciences
https://doi.org/10.1007/s10072-021-05347-3

ORIGINAL ARTICLE

Diagnosis and treatment of Chiari malformation and syringomyelia in adults: international consensus document

Palma Ciaramitaro[1,2] · Luca Massimi[3] · Alessandro Bertuccio[4] · Alessandra Solari[5] · Mariangela Farinotti[5] · Paola Peretta[6] · Veronica Saletti[7] · Luisa Chiapparini[8] · Andrea Barbanera[4] · Diego Garbossa[1] · Laura Valentini[9] · On behalf of the International Experts Jury of the Chiari Syringomyelia Consensus Conference, Milan, November 11-13, 2019

Received: 15 January 2021 / Accepted: 21 May 2021

Abstract

Background Syringomyelia and Chiari malformation are classified as rare diseases on Orphanet, but international guidelines o diagnostic criteria and case definition are missing. Aim of the study: to reach a consensus among international experts o controversial issues in diagnosis and treatment of Chiari 1 malformation and syringomyelia in adults.

Methods A multidisciplinary panel of the Chiari and Syringomyelia Consortium (4 neurosurgeons, 2 neurologists, 1 neuroradi ologist, 1 pediatric neurologist) appointed an international Jury of experts to elaborate a consensus document. After an evidence based review and further discussions, 63 draft statements grouped in 4 domains (definition and classification/planning/surgery isolated syringomyelia) were formulated. A Jury of 32 experts in the field of diagnosis and treatment of Chiari and syringomyeli and patient representatives were invited to take part in a three-round Delphi process. The Jury received a structured questionnair containing the 63 statements, each to be voted on a 4-point Likert-type scale and commented. Statements with agreement <75% were revised and entered round 2. Round 3 was face-to-face, during the Chiari Consensus Conference (Milan, November 2019

Results Thirty-one out of 32 Jury members (6 neurologists, 4 neuroradiologists, 19 neurosurgeons, and 2 patient associatio representatives) participated in the consensus. After round 2, a consensus was reached on 57/63 statements (90.5%). The si difficult statements were revised and voted in round 3, and the whole set of statements was further discussed and approved.

Conclusions The consensus document consists of 63 statements which benefited from expert discussion and fine-tuning, servin clinicians and researchers following adults with Chiari and syringomyelia.

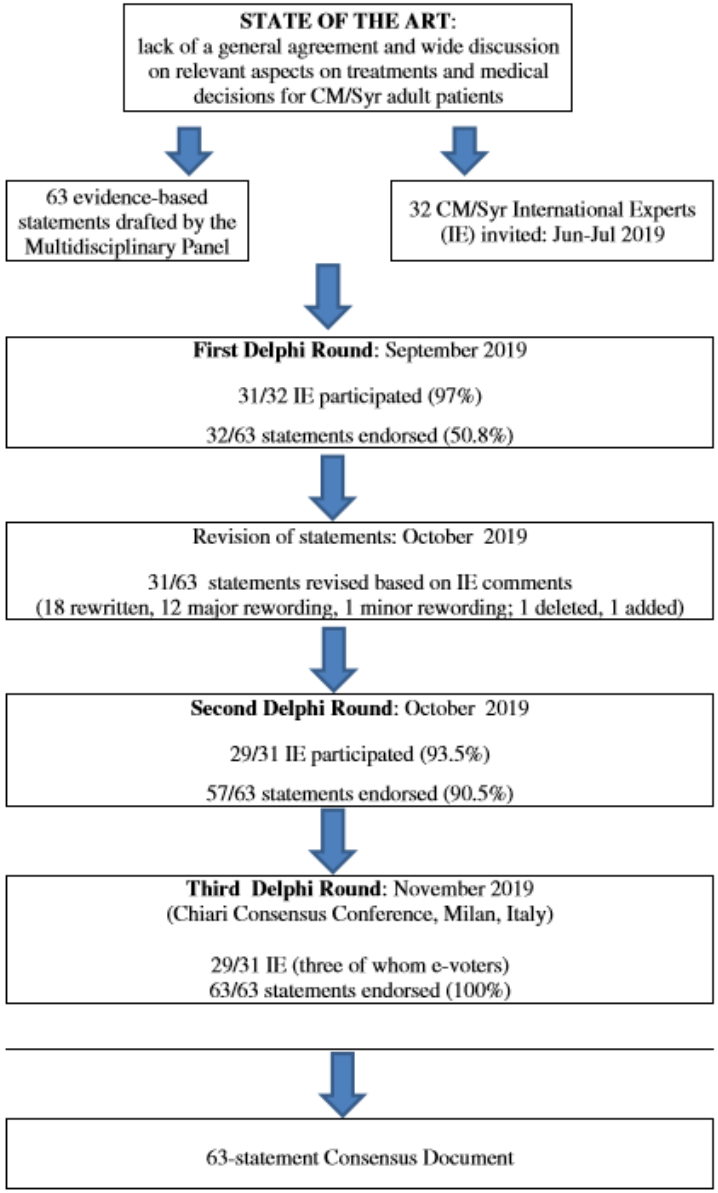


Table 1 Classifications and definitions

Chiari malformation classification

C1

CM1: paraxial mesoderm disorder, with abnormalities of the posterior cranial fossa (mostly small) and the consequent herniation of the cerebellar tonsils:
CM1-A: with syringomyelia on MRI

CM1-B: without syringomyelia on MRI

Agreement: 85.4%

Grade D

C2

CM2: due to prenatal CSF hypotension always associated with myelomeningocele and, in some cases, with hydrocephalus and syringomyelia. Other types of intracranial defects (hypoplastic tentorium cerebelli, cranial lacunae, anomalies of the Sylvius aqueduct) may coexist (1).

Agreement: 85.4%

Grade D

C3

CM-1.5: cerebellar tonsils and brainstem herniation (kinking) below the McRae line; defined "complex" if also skeletal deformities of cranio-vertebral junction and cervical spine (Klippel-Feil anomaly, atlanto-occipital fusion, basilar invagination, retroversion of the odontoid process) are present.

Agreement: 81.3%

Grade D

C4

CM-0: obliteration of the cisterna magna (due to arachnoid adhesions) and/or volumetrically small posterior fossa, with cerebellar tonsils positioned at the foramen magnum and a syringomyelia in the cervical spinal cord.

Agreement: 81.3%

Grade D

C5

CM1 should be differentiated from cerebellar tonsil herniation secondary to space-occupying lesions (hydrocephalus, arachnoid cysts, brain tumors) and termed "acquired tonsillar ectopia."

Agreement: 95.8%

Grade D

Definitions

D1

Chiari 1 malformation

One or both cerebellar tonsil cauda

< 3 mm, normal

≥ 5 mm, CM1

3–5 mm: this condition is definitel

• Syrinx

• Peg-like deformation of the tons

Agreement: 83.7%

Grade D

D2

Chiari syndrome

Chiari syndrome (CS) is the clinica

defined."

Clinical diagnostic criteria (symptoms and neurological signs) are:

a) Headache caused by Chiari type I malformation, usually occipital or suboccipital, of short duration (less than five minutes) and provoked/precipitated by cough or other Valsalva-like maneuvers

b) Symptoms and/or signs of brainstem (i.e., nystagmus, dysfagia, sleep apnea), cerebellar (ataxia), and/or cervical cord dysfunction (i.e., muscles hypotrophy, sensory and motor deficits)

c) Otoneurological symptoms and/or signs (e.g., dizziness, disequilibrium, sensations of alteration in ear pressure, hypoacusia or hyperacusia,

Chiari 1 malformation: differential diagnosis

DD1

False tonsil descent due to intracranial hypotension must be excluded by clinical pattern, MRI pattern, and contrast-enhanced MRI.

Agreement: 85.2%

Grade D

DD2

False tonsil descent due to intracranial hypertension must be excluded by clinical pattern, fundoscopy, and MRI pattern, completed with venous angio-MRI; direct ICP monitoring could be indicated in selected cases.

Agreement: 96.3%

Grade D

Table 1 Planning for CM1 in children: indications to surgery

1	In *asymptomatic* children with incidentally discovered, isolated CM1 and *no syringomyelia*, surgery is not indicated	Agreement: 94.1%
2	In asymptomatic children with incidentally discovered CM1 and syringomyelia, surgery is indicated in cases of *syrinx larger than 5–6 mm*, and smaller syrinx increasing in size	Agreement: 82.4%
3	In children with *epilepsy* and CM1, surgical treatment of CM1 does not improve the seizure disorder	Agreement: 94.1%
4	In children with CM1 and *cognitive* and/or *behavioral* disorders (such as autism) not able to correctly report their symptoms because of communication impairment, careful clinical and instrumental assessments are mandatory to detect symptoms and signs of cerebellar tonsils ectopia	Agreement: 97.1%
5	In children with CM1 and cognitive and/or behavioral disorders (such as autism) and no clear CM1 symptoms, surgery is not indicated to improve the clinical picture	Agreement: 91.2%
6	Surgical treatment is not decided on *MRI pattern* in asymptomatic children with tonsillar ectopia ≥ 20 mm without syringomyelia	Agreement: 88.2%

Neurological Sciences (2022) 43:1311–1326
https://doi.org/10.1007/s10072-021-05317-9

ORIGINAL ARTICLE

Diagnosis and treatment of Chiari malformation type 1 in children: the International Consensus Document

Luca Massimi[1] · Paola Peretta[2] · Alessandra Erbetta[3] · Alessandra Solari[4] · Mariangela Farinotti[4] · Palma Ciaramitaro[5] · Veronica Saletti[6] · Massimo Caldarelli[1] · Alexandre Casagrande Canheu[7] · Carlo Celada[8] · Luisa Chiapparini[3] · Daniela Chieffo[9] · Giuseppe Cinalli[10] · Federico Di Rocco[11] · Marika Furlanetto[12] · Flavio Giordano[13] · George Jallo[14] · Syril James[15] · Paola Lanteri[16] · Christian Lemarchand[17] · Martina Messing-Jünger[18] · Cecilia Parazzini[19] · Giovanna Paternoster[15] · Gianluca Piatelli[20] · Maria. A. Poca[21] · Prab Prabahkar[22] · Federica Ricci[23] · Andrea Righini[19] · Francesco Sala[24] · Juan Sahuquillo[21] · Marcus Stoodley[25] · Giuseppe Talamonti[26] · Dominic Thompson[27] · Fabio Triulzi[28] · Mino Zucchelli[29] · Laura Valentini[12] · International Experts - Jury of the Chiari & Syringomyelia Consensus Conference, "Milan, November 11–13, 2019"

Chiari1 Malformation and Epilepsy in children: a missing relationship.
L. Massimi et al. J.Clin.Med., ***2022****,11 (20), 6182*

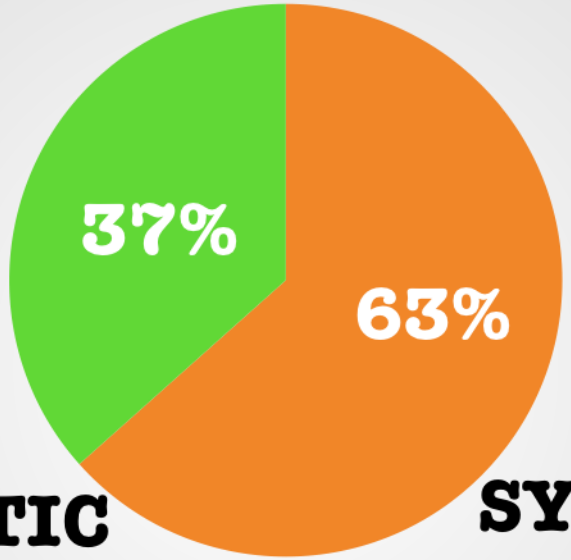
Table 2 Planning for CM1 in adults: surgical indications and follow-up

1	In symptomatic CM1 without syringomyelia, surgery is indicated in adults with headache (typical, invalidating, and resistant to therapy) and auditory/cerebellar/bulbar/ spinal signs at neurological examination.	Agreement: 100% Grade B
2	In asymptomatic CM1 without syringomyelia, surgery is not indicated.	Agreement: 100% Grade B
3	In asymptomatic CM1 without syringomyelia, a follow-up is mandatory with neurological evaluation and whole neuraxis MRI with a schedule based on clinical and MRI picture.	Agreement: 81.5% Grade C
4	In CM1 with syringomyelia, surgery is indicated for holocord syringomyelia, clinical/MRI worsening, central syringe and Vaquero Index >0.5 or eccentric syringe, syringomyelia-syringobulbia with spinal/bulbar signs	Agreement: 96.3% Grade B
5	In asymptomatic CM1 with syringomyelia, SEPs, BAEPs, and MEPs may be performed to evidence subclinical deficits, but surgical indications are based on clinical and neuro-radiological grounds.	Agreement: 92.3% Grade C
6	In CM1 with syringomyelia, polysomnography is indicated in case of reported/suspected sleep apneas.	Agreement: 92.6% Grade D
7	In asymptomatic CM1 without syringomyelia, neurophysiological study by SEPs-BAEPs-MEPs may be performed to evidence subclinical deficits, but surgical indications are based on clinical and neuro-radiological grounds.	Agreement: 85.2% Grade C
8	In CM1 without syringomyelia, polysomnography is indicated in case of reported/suspected sleep apneas.	Agreement: 88.9% Grade D

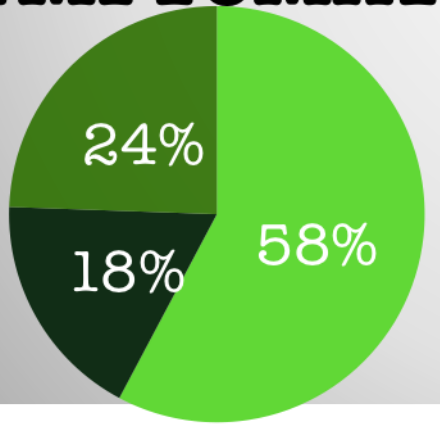
NATURAL HISTORY (FU≥10 yrs)

CRESSC series* (2008 to 2024)

*sample of 100 pts
(86 F; 37 M; mean age
55 ± 14, range 22-83)

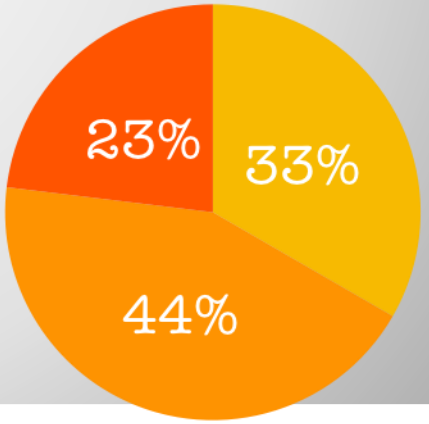


ASYMPTOMATIC



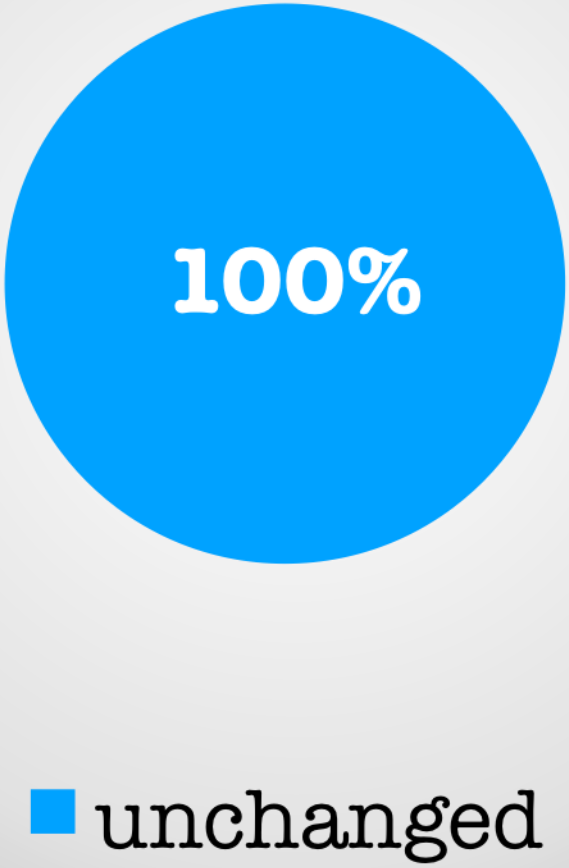
CM1 CM1+Syr Syr/Hydro

SYMPTOMATIC

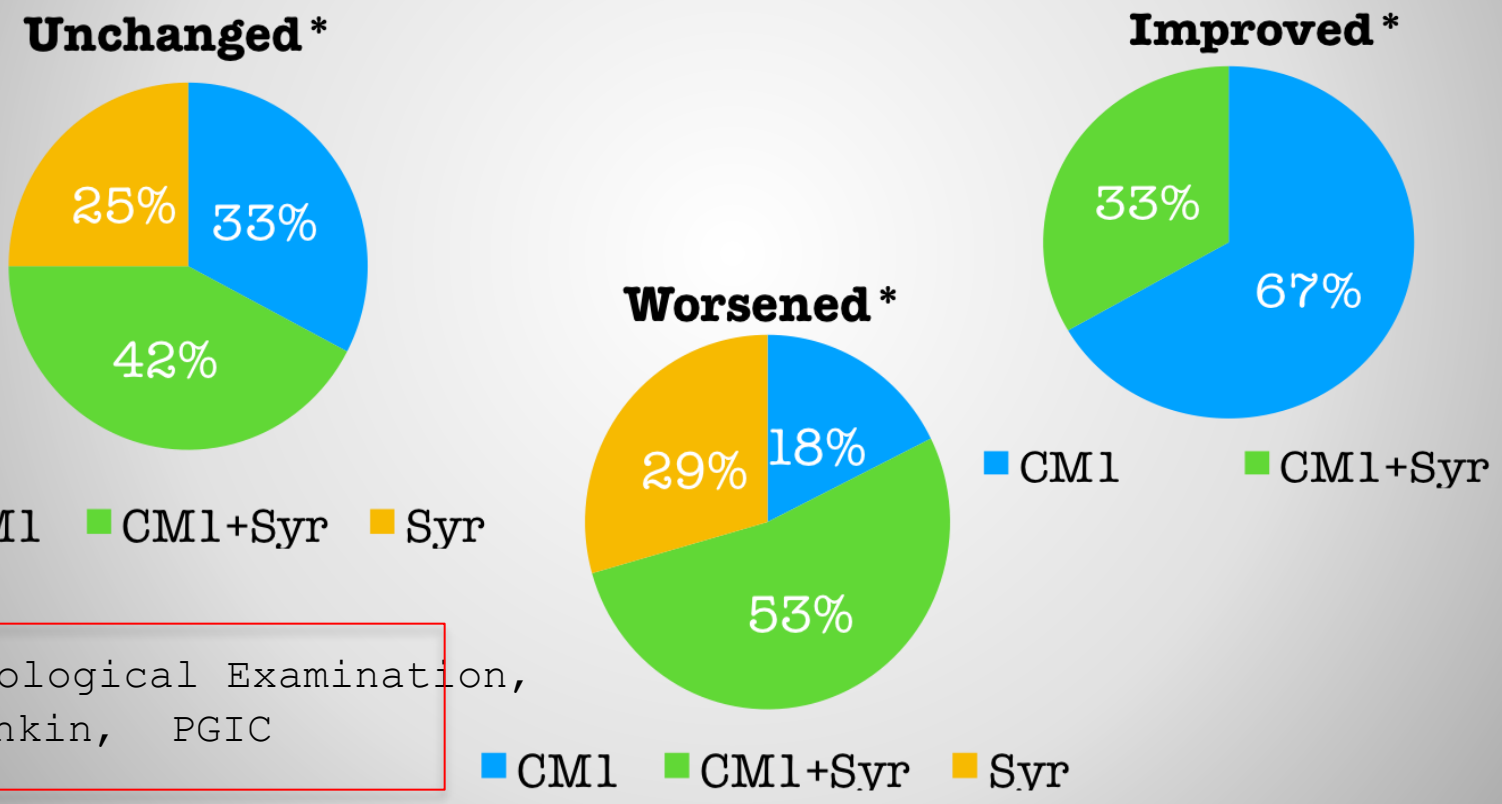


CM1 CM1+Syr Syr/Hydro

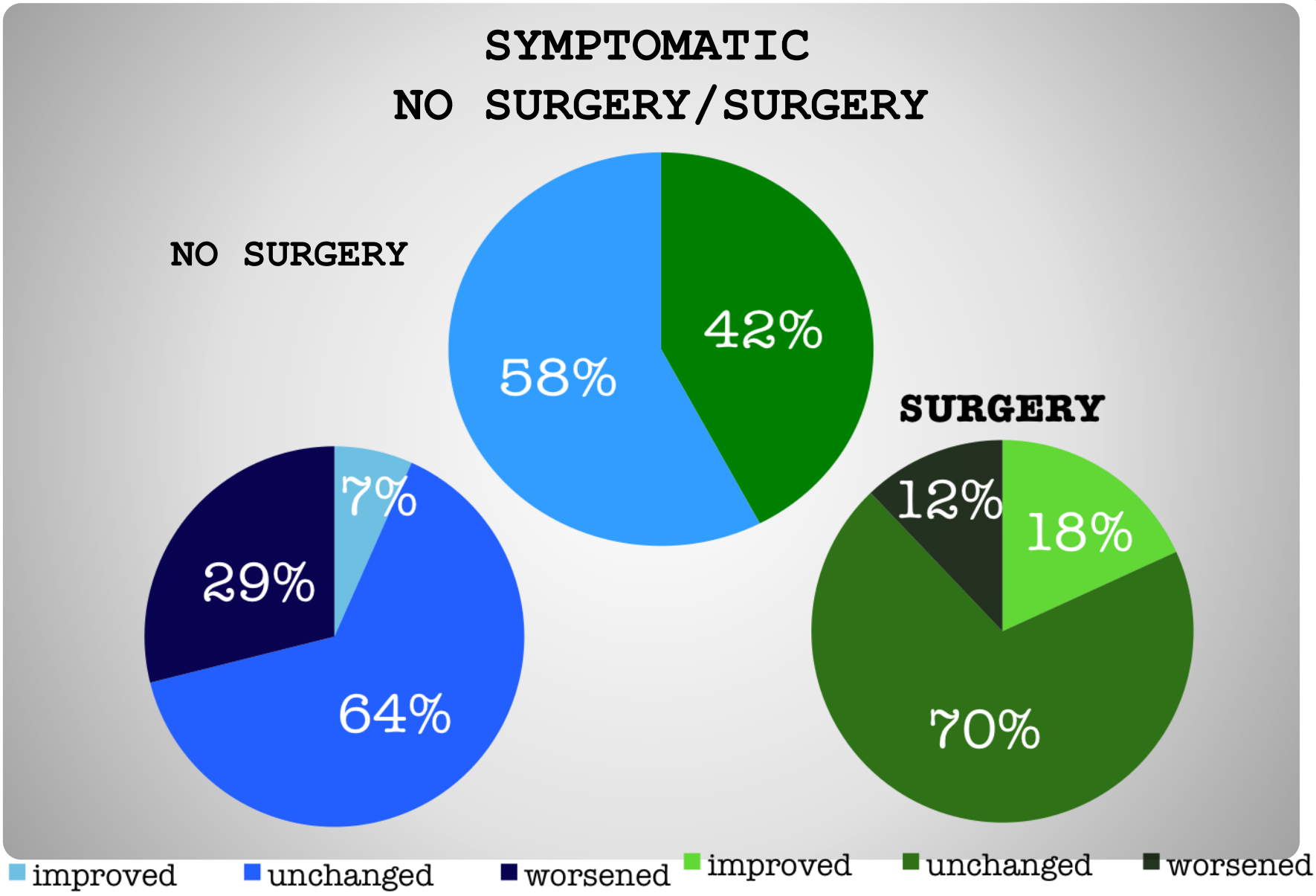
ASYMPTOMATIC NATURAL HISTORY (≥10 yrs FU=37 pts)



SYMPTOMATIC NATURAL HISTORY (≥10 yrs neurological FU=63 pts)



*Neurological Examination, Rankin, PGIC


SYMPTOMATIC
NO SURGERY/SURGERY
NO SURGERY
58%
42%
SURGERY
12%
18%
70%
7%
29%
64%
improved
unchanged
worsened
improved
unchanged
worsened

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