UPPER CERVICAL MYELOPATHY WITH METATROPIC DYSPLASIA

REPORT OF 4 SURGICAL CASES

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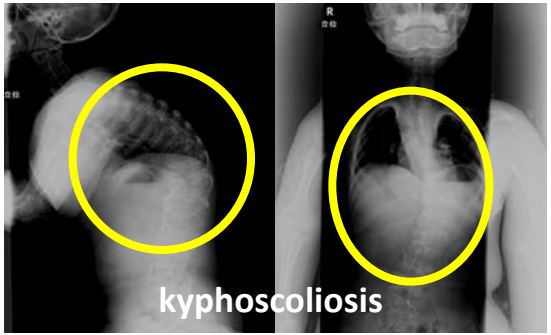
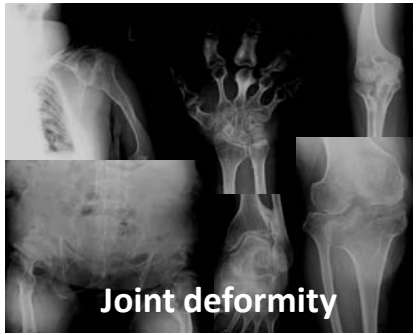
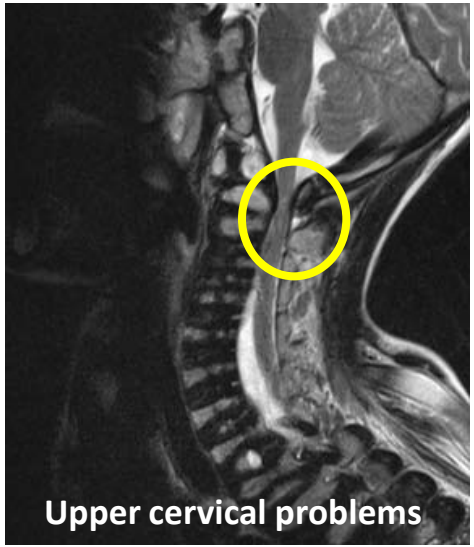
INTRODUCTION

Metatropic Dysplasia (=MD) is a very rare skeletal dysplasia presenting dwarfism, which is characterized by **short limbs at birth** and **rapid collapse of thoracolumbar spine into kyphoscoliosis during infancy**. It is also one of the **TPRV4 gene** related disease family.







Upper cervical myelopathy based on hypoplasia of odontoid process and/or lax ligaments is known as serious complication in MD patients.

But there have been few reports on its clinical feature including surgical treatment.

PURPOSE

To evaluate the clinical features and the surgical outcomes of upper cervical myelopathy in MD patients.

MATERIALS

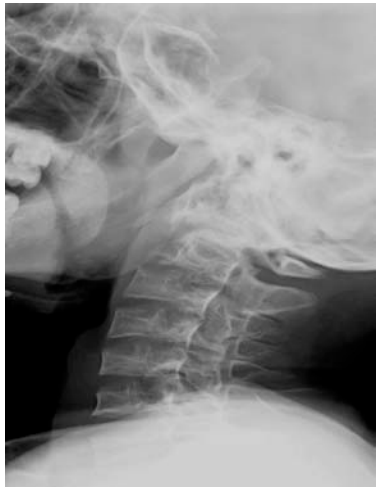
4 surgical cases of upper cervical myelopathy in MD

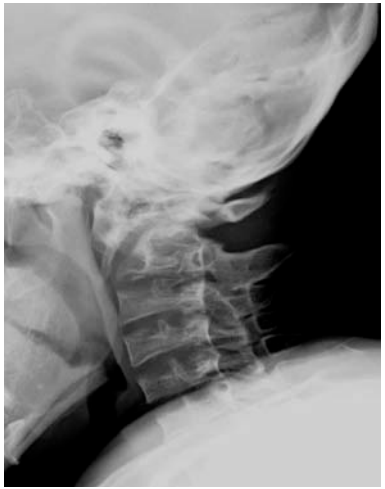
- # Surgically treated during 2005 to 2011
- # Sex : 3 males and 1 female
- # The ages at surgery : 6, 13, 15 and 44 years old
- # Postoperative follow-up period : 42.5 months (range 7 – 69)

METHODS

Retrospective analysis of :

1) Preoperative Clinical Features
2) Complications
3) Radiological Findings
4) Surgical Procedures
5) Intra/Postoperative Complications
6) Surgical Outcomes





RESULTS 1

Preoperative Clinical Features

- All 4 cases presented progressive spastic tetraplegia with or without respiratory dysfunction
- Accurate evaluation of neurological dysfunction was difficult because of existing complications (mental retardation, severe lower limb joint deformity and restrictive respiratory dysfunction)

◆ Upper limb function

Unable to use chopsticks but able to eat with a spoon	3 cases
Unable to eat without support	1 case

◆ Lower limb function

Unable to stand nor walk without support	4 cases

◆ Respiratory function

Severe respiratory dysfunction which needs NIPPV *	2 cases
Almost normal	2 cases

*Non-invasive Positive Pressure Ventilation

RESULTS 2

Complications

- Slight mental retardation in 2 cases
- Severe lower limb joint deformity in 2 cases
- Severe restrictive respiratory dysfunction with hypoplastic thorax in 2 cases*

*Both of which needed NIPPV (Non-invasive Positive Pressure Ventilation)

Radiological Findings

- All 4 cases presented severe canal stenosis at C1/2, which were caused by the following pathologies:
  - Atlantoaxial subluxation with hypoplasia of odontoid process in 3 cases
  - Hyperostosis of odontoid process in 1 case
- **Atlantoaxial instability** was **apparently present** in 2 cases
  - **suspicious** in 1 case
  - **not present** in 1 case

RESULTS 3

Surgical Procedure

C1/2 instability (-) → Posterior decompression only 1 case

C1/2 instability (+) or suspicious → Posterior decompression & fusion (including occipital bone) 3 cases

Case	Age at operation	Sex	Hypoplasia of odontoid process	C1/2 Subluxation	C1/2 Instability	Range of Stenosis	Decompression	Fusion	Instrument
1	6	f	+	+	−	C1-7	Oc-C7	-	-
2	13	m	+	+	+	C1-3	Oc-C4	Oc-C2	-
3	15	m	+	+	+	C1/2	Oc-C1/2	Oc-C2	+
4	44	m	Unknown (Odontoid hyperplasia)	Unknown	Suspicious (micro)	C1/2	C1-3	Oc-Th3	+

RESULTS 4

Intra/Postoperative Complication

No major complication occurred in this series

Surgical Outcomes

- Upper limb function

  Apparent improvement in all cases , such as feeding activity, writing , drawing and handling electric wheel chair

- Lower limb function:

  Independent gait was achieved in 2 cases

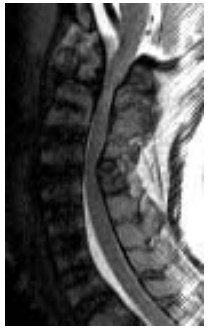
  Elongation of the distance of supported gait was achieved in 1case

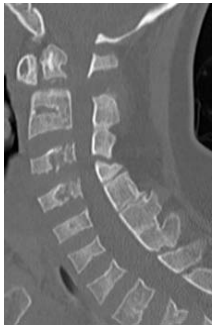
  *No remarkable change in 1 case with severe lower limb joint deformity who had been using wheel chair for approximately 30 years

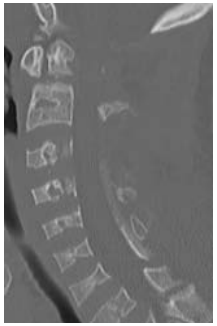
- Respiratory function:

  In 1 case with respiratory dysfunction, $PaCO_2$ remarkably improved from 80mmHg to 50mmHg

CASE 1 (6 y.o. female)


Preop.MRI


Preop.CT


Postop.CT

Neurology: Progressive tetraplegia

ADL: Unable to stand without support nor use chopsticks but able to eat with spoon

Complication: Slight mental retardation(+)

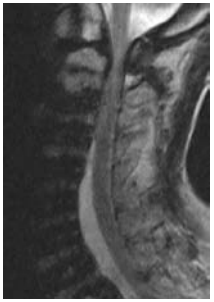
Radiology: C1/2 stenosis (+) Hypoplasia of odontoid process (+) C1/2 instability (-)

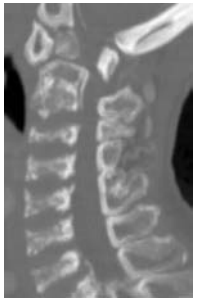
Surgical Procedure: C1-C7 decompression (C1 posterior arch resection & C2-7 laminoplasty)

Outcome: Independent gait

Improvement in coordinated motion such as feeding activity with spoon & folk

CASE 2 (13 y.o. male)


Preop.MRI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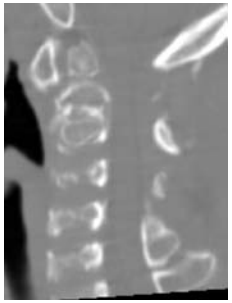

Postop.CT

Neurology: Progressive tetraplegia with respiratory dysfunction

ADL: Unable to stand without support nor use chopsticks but able to eat with spoon

Complication: Severe lower limb joint deformity (Hip & Knee) ,

Respiratory dysfunction (with NIPPV support)

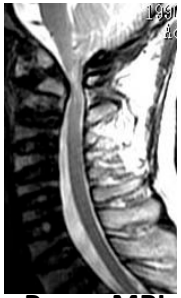
Radiology: C1/2 stenosis (+) Hypoplasia of odontoid process (+) Instability (+)

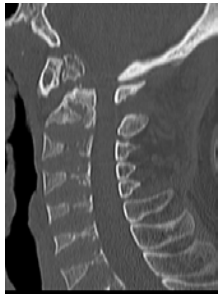
Surgical Procedure: C1-4 decompression & fusion without instrumentation

Outcomes: Increase in distance of supported gait

Improvement in coordinated motion such as feeding activity and writing

CASE 3 (15 y.o. male)


Preop.MRI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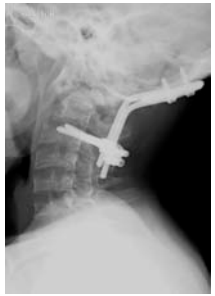

Postop.Xp

Neurology: Progressive tetraplegia

ADL: Unable to stand up without support nor eat without support

Complication: Slight mental retardation

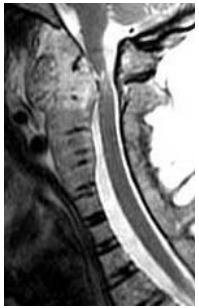
Radiology: C1/2 stenosis (+) Hypoplasia of odontoid process (+) Instability (+)

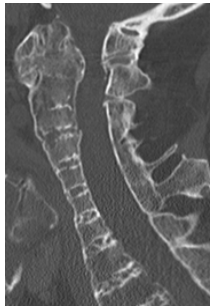
Surgical Procedure: Oc-C1decompression & Oc-C3 fusion with instrumentation

Outcomes: Independent Gait

Improvement in coordinated motion such as feeding activity, writing and drawing

CASE 4 (44 y.o. male)


Preop.MRI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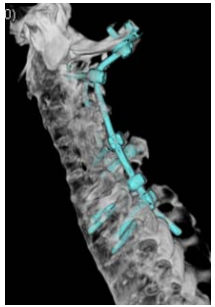
Postop.CT

Neurology: Progressive tetraplegia with respiratory dysfunction

ADL: Unable to stand without support nor use chopsticks but able to eat with spoon

Complication: Severe lower limb joint deformity (using wheel chair for a long time)

Respiratory dysfunction (with NIPPV support)

Radiology: C1/2 stenosis (+) Hyperostosis of odontoid process (+)

Bony ankylosis of whole spine (+) Micro instability at C1/2 was suspicious

Surgical Procedure: C1-3 decompression & Oc-Th3 fusion with instrumentation

Outcomes: Improvement in coordinated motion such as feeding activity with spoon & folk and handling of electric wheel chair

Improvement in $PaCO_2$ from 80mmHg to 50mmHg

CONCLUSION

- Evaluation of accurate neurological dysfunction was difficult in MD patients with upper cervical myelopathy , because of other complications (such as mental retardation, joint deformity and restrictive respiratory dysfunction.)
- Preoperative symptoms of myelopathy tend to be severe, which were unable to walk nor stand without support in all cases.
- Canal stenosis and spinal cord compression at C1/2 presented in all cases , but the pathologies of which were varied in the respective cases.
- With special attention to respiratory dysfunction, surgical treatment was effective and the outcomes were satisfactory.