

## STUDY OSTEOPETROSIS - Status and Follow Up

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Patients Initials \_\_\_\_\_ Sex \_\_\_\_\_ Date of birth \_\_\_\_\_ Institution \_\_\_\_\_ Local ID \_\_\_\_\_  
 (last name/first name) (m=1, f=2) (DD.MM.YYYY)

Date of examination: \_\_\_\_\_ (DD.MM.YYYY) Follow up after: \_\_\_\_\_ years Status: \_\_\_\_\_  
 (1= transplanted, 2= not transplanted, 3= unknown)

**!! Please make sure that the consent form is signed before transmitting these data !!**

### PHYSICAL STATUS

- 1. Vision:**
- 1  no optical atrophy, and/or presence of VEP, and/or eye fixation  
 2  partial optical atrophy, and/or reduced VEP response, and/or presence of eye fixation, and/or nystagmus  
 3  total optical atrophy, and/or absence of VEP, and/or amaurotic nystagmus and/or no eye fixation  
 4  unknown
- 2. Hearing:**
- 1  normal hearing  
 2  defective hearing, tested with: \_\_\_\_\_  
 3  deaf  
 4  unknown
- 3. Height:**
- 1  > 10. percentile  
 2  > 3. and < 10. percentile  
 3  < 3. percentile  
 4  unknown
- 4. Weight:**
- 1  > 10. percentile  
 2  > 3. and < 10. percentile  
 3  < 3. percentile  
 4  unknown
- 5. Head Circumference:**
- 1  > 97. percentile  
 2  90. - 97. percentile  
 3  10. - 90. percentile  
 4  3. - 10. percentile  
 5  < 3. percentile  
 6  unknown
- 6. Dentition:**
- 1  normal  
 2  extra teeth  
 3  worse than normal, delayed dentition  
 4  very bad, surgery necessary  
 5  unknown
- 7. Other clinical indications for osteopetrosis:**
- |  | no                       | yes                      | unknown                  |
|--|--------------------------|--------------------------|--------------------------|
| a hepatomegaly                         | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| b splenomegaly                         | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| c breathing problems                   | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| d seizures                             | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| e fractures                            | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| f exophthalmos                         | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| g frontal bossing / prominent forehead | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| h osteomyelitis                        | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

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<input type="text"/>	<input type="text"/> (m=1, f=2)	<input type="text"/> . <input type="text"/> . <input type="text"/>	<input type="text"/>	<input type="text"/>
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**MENTAL STATUS**

- 8. Neurological Examination:**
- 1  normal
- 2  mildly abnormal (minor neurological signs as broad gait, dysdiadochokinesis, dysmetria)
- 3  severely abnormal (paresis, spasticity, cerebral nerve palsy, ataxia)
- 4  not done

- 9. Bayley II:**
- 1  85 - 114: normal performance
- 2  70 - 84 : mildly delayed performance
- 3  50 - 69 : moderately delayed performance
- 4  < 50 : severely delayed performance
- 5  not done

**Children with severe visual handicap / blindness:**

Bayley II mental scale: use only language items and calculate mental development index (MDI) by dividing passed language items by expected language items for corrected age.

**10. Clinical Impression (if Bayley - MDI is not done or not applicable):**

- 1  normal
- 2  mild retardation = developmental age estimated 15 - 30% below age
- 3  moderate retardation = developmental age estimated 31 - 50% below age
- 4  severe retardation = developmental age estimated > 50% below age
- 5  unknown

**11. Autism - Pervasive developmental disorder - not otherwise specified (PDD-NOS):**

**A** = impairment of social interaction

**B** = impairment of communication

**C** = restricted repetitive stereotyped behavior patterns

**D** = delays in social interaction or language as used in social communication or symbolic / imaginary play before age 3 years

- 1  normal (no behavior type out of A-D)
- 2  mild (1 out of A-D)
- 3  moderate (2 or 3 out of A-D)
- 4  severe (all out of A-D)
- 5  not assessed / impossible to assess

- 12. School attendance:**
- 1  regular school / kindergarten
- 2  special school / kindergarten
- 3  no school / kindergarten
- 4  hospitalized
- 5  unknown
- 6  not applicable (age)

- 13. Quality of life (parents / carers view):**  **Visual analogue scale 0 - 10 ( 0 = very bad, 10 = excellent )**

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**CLINICAL FINDINGS**

**14. Haematological status:**

	<b>no</b>	<b>yes</b>	<b>unknown</b>
a leukocytosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
b leukocytopenia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
c thrombocytopenia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
d anemia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
e transfusion dependant - platelets	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
f transfusion dependant - red cells	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**15. Calcium metabolism:**

1 <input type="checkbox"/> normal	→		<b>no</b>	<b>yes</b>	<b>unknown</b>
2 <input type="checkbox"/> pathological		a hypocalcemia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3 <input type="checkbox"/> not done		b hypercalcemia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4 <input type="checkbox"/> unknown		c nephrocalcinosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
		d other, specify: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**16. Radiologic indicators of osteopetrosis:**

1 <input type="checkbox"/> normal	→		<b>no</b>	<b>yes</b>	<b>unknown</b>
2 <input type="checkbox"/> pathological		a bone-in-bone	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3 <input type="checkbox"/> not done		b craniosynostosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4 <input type="checkbox"/> unknown		c increased general skeletal sclerosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
		d metaphyseal widening	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
		e other, specify: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**17. Bone marrow histology:**

1 <input type="checkbox"/> normal	→		<b>no</b>	<b>yes</b>	<b>unknown</b>
2 <input type="checkbox"/> pathological		a osteosclerosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3 <input type="checkbox"/> not done		b normal osteoclasts	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4 <input type="checkbox"/> unknown		c decreased osteoclasts	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
		d other, specify: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**18. Magnetic Resonance Imaging (MRI / CT):**

1 <input type="checkbox"/> normal	→		<b>no</b>	<b>yes</b>	<b>unknown</b>
2 <input type="checkbox"/> pathological		a hydrocephalus	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3 <input type="checkbox"/> not done		b narrowed foramina	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4 <input type="checkbox"/> unknown		c other, specify: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**19. Chimerism:**

1  not done / not applicable  
 2  done  
 3  unknown

PB/BM	Date of examination	If sorted, type of cells	% of donor cells	Method (FISH, STR, other)	Laboratory (city)	Treatment				
						Immunosuppression: none	unchanged	increased	reduced	stopped
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
_____	_____	_____	_____	_____	_____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

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**SURVIVAL STATUS**

**20. Performance Status Criteria:**

Karnofsky / Lansky	Karnofsky Description (Patients elder than 16 years)	Lansky Description (Patients younger than 16 years)
100 <input type="checkbox"/>	Normal, no complaints, no evidence of disease.	Fully active, normal.
90 <input type="checkbox"/>	Able to carry on normal activity; minor signs or symptoms of disease.	Minor restrictions in physically strenuous activity.
80 <input type="checkbox"/>	Normal activity with effort; some signs or symptoms of disease.	Active, but tires more quickly.
70 <input type="checkbox"/>	Cares for self, unable to carry on normal activity or do active work.	Both greater restriction of and less time spent in play activity.
60 <input type="checkbox"/>	Requires occasional assistance, but is able to care for most of his/her needs	Up and around, but minimal active play, keeps busy with quieter activities.
50 <input type="checkbox"/>	Requires considerable assistance and frequent medical care.	Gets dressed, but lies around much of the day; no active play; able to participate in all quiet play and activities.
40 <input type="checkbox"/>	Disabled, requires special care and assistance.	Mostly in bed; participates in quiet activities.
30 <input type="checkbox"/>	Severely disabled, hospitalization indicated. Death not imminent.	In bed; needs assistance even for quiet play.
20 <input type="checkbox"/>	Very sick, hospitalization indicated. Death not imminent.	Often sleeping; play entirely limited to very passive activities.
10 <input type="checkbox"/>	Moribund, fatal processes progressing rapidly.	No play; does not get out of bed.

**21. Survival:**

1  alive free of disease

2  alive with disease

3  died

4  lost to follow-up / unknown

**22. Treatment:**

1  transplantation in preparation

2  supportive therapy

3  other therapy:

	no	yes	unknown
a calcitriol	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
b corticosteroids	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
c IFN-γ	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

4  unknown

**23. Date of death:**

.    .     (DD.MM.YYYY)

Main Cause of death:

progression of disease

transplantation related cause: (tick all that apply)

GvHD

graft failure

pulmonary toxicity

cardiac toxicity

infection

VOD

posttransplant lymphoproliferative disorder

other: \_\_\_\_\_

other: \_\_\_\_\_

unknown

**24. Comment:**

Date

Clinic Stamp

Name

Signature