

**STUDY OSTEOPETROSIS - Registration and Genetics**

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

Date of examination: \_\_\_\_\_ Institution: \_\_\_\_\_

**!! Please make sure that the consent form is signed before transmitting these data !!** Date of informed consent: \_\_\_\_\_

**PATIENT BACKGROUND**

- 1. Ethnic background:**
- 1  European (Central/Northern/Western/Eastern)
  - 2  Mediterranean
  - 3  Arabian / Middle East
  - 4  other, specify: \_\_\_\_\_
  - 5  unknown

- 2. Family History:**
- 1  negative
  - 2  positive, please specify: \_\_\_\_\_
  - 3  unknown

- 3. Consanguinity:**
- 1  no
  - 2  yes
  - 3  unknown

**GENETICS**

- 4. DNA analysis:**
- 1  not done
  - 2  done
  - 3  unknown

If done, please specify:

5. Gene mutation:	no	yes	not done	unknown
a TCIRG1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
b CLCN7	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
c OSTM1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
d RANKL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
e RANK	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
f SNX10	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
g KINDLIN/FERMT3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
h other: _____	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**6. Type of mutation:**

- 1  homozygous
- 2  compound heterozygous
- 3  heterozygous
- 4  unknown

Patients Initials [ ][ ]	Sex [ ] (m=1, f=2)	Date of birth [ ][ ] . [ ][ ] . [ ][ ][ ][ ] (DD.MM.YYYY)	Clinic _____	Local ID [ ][ ][ ][ ][ ][ ][ ][ ]
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**7. Consequence Type:**

	no	yes	unknown
a single AA change	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
b major insertion / deletion	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
c frameshift / stop codon	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
d splice site mutation	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**8. Specify:**

a position / nucleotide > nucleotide: \_\_\_\_\_  
 \_\_\_\_\_

b position / aminoacid > aminoacid: \_\_\_\_\_  
 \_\_\_\_\_

**9. Genetic Laboratory:**

1  Milan  
 2  Paris  
 3  Ulm  
 4  other, specify: \_\_\_\_\_  
 5  unknown

**!! Please add "Status and Follow Up" as well as "Transplantation" form, if applicable !!**

Forms added: 1  Transplantation  
 2  Status and Follow Up

\_\_\_\_\_ Date

\_\_\_\_\_ Clinic Stamp

\_\_\_\_\_ Name

\_\_\_\_\_ Signature