

National Marrow Donor Program®
**Insert XIV – Mucopolysaccharidoses
 and Other Storage Diseases**

Registry Use Only

Sequence
 Number:

Date
 Received:

Unrelated

Recipient NMDP ID: - -

Recipient Last Name:

Recipient Local ID (optional):

Today's Date: / / TC Code:

Month Day Year

Date of Transplant for which this form is being completed: / /

Month Day Year

Product type: Marrow (Form 120) PBSC (Form 520) Cord blood (Form 620)

This form must be accompanied by Form 120, 520, 620 – Recipient Baseline and Transplant Data. All information in the box above, including the date, should be identical with the corresponding Form 120, 520, 620. Information should come from an actual examination by the Transplant Center physician, or the physician who followed the recipient post-transplant, or abstraction of the recipient's medical records.

1. Which enzyme deficiency was detected at diagnosis?

Mucopolysaccharidosis

- 1 α -L-iduronidase (Hurler – MPS I)
- 2 Iduronate sulfatase (Hunter – MPS II)
- 3 Heparan N-sulfatase (Sanfilippo A – MPS IIIA)
- 4 α -N-acetylglucosaminidase (Sanfilippo B – MPS II B)
- 5 Acetyl CoA: α -glucosaminide acetyltransferase (Sanfilippo C – MPS IIIC)
- 6 N-acetylglucosamine 6-sulfatase (Sanfilippo D – MPS IIID)
- 7 Galactose 6-sulfatase (Morquio A – MPS IVA)
- 8 β -galactosidase (Morquio B – MPS IVB)
- 9 N-acetyl galactosamine 4-sulfatase (Maroteaux-Lamy – MPS VI)
- 10 β -glucuronidase (Sly syndrome – MPS VII)

Other Storage Diseases

- 11 Glucocerebrosidase (Gaucher)
- 12 Acid sphingomyelinase (Niemann-Pick)
- 13 Phosphotransferase (Mucopolipidosis II or I-cell)
- 14 Acid lipase (Wolman)
- 15 α -fucosidase (Fucosidosis)
- 16 Neuronal ceroid-lipofuscinosis enzyme – NCL 1 (infantile): PPT-palmitoyl protein thioesterase
- 17 Neuronal ceroid-lipofuscinosis enzyme – NCL 2 (classic late infantile): transpeptidase
- 18 α - or β -mannosidase (Mannosidosis)
- 19 Aspartyl glucosaminidase (Aspartylglucosaminuria)
- 20 Hypoxanthine-guanine phosphoribosyltransferase (Lesch-Nyhan)
- 21 Other storage disease, specify: _____

2. Record the leukocyte enzyme levels at diagnosis:

- a. Patient enzyme level: / • 1 nmol/hr/mg protein 2 pmol/hr/mg protein
- b. Donor enzyme level: / • 1 nmol/hr/mg protein 2 pmol/hr/mg protein

Recipient NMDP ID: - -

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3. Was treatment given for the disease between diagnosis and transplant?

- 1 yes →
2 no
3 unknown

4. Specify:

- a. Enzyme replacement 1 yes 2 no 3 unknown
b. Substrate deprivation/inhibitor 1 yes 2 no 3 unknown
c. Gene transfer/gene therapy 1 yes 2 no 3 unknown

Clinical Status Pre-Transplant

5. Was cerebrospinal fluid (CSF) testing done pre-transplant?

- 1 yes →
2 no
3 unknown

6. Report results of most recent tests:

- a. Opening pressure cm H₂O
1 yes →
2 no
3 unknown
- b. Total protein . mg/dL 2 g/L
1 yes →
2 no
3 unknown
- c. **Serum** albumin . mg/dL 2 g/L
1 yes →
2 no
3 unknown
- d. **Serum** IgG . mg/dL 2 g/L
1 yes →
2 no
3 unknown

7. Date of most recent test:

/ /
Month Day Year

8. Magnetic Resonance Imaging (MRI) of the brain/spine pre-transplant:

- 1 yes →
2 no
3 unknown

9. Specify location of abnormalities:

(If possible, attach a copy of the report.)

a. Ventricular (hydrocephalus):

- 1 yes
2 no
3 unknown

b. Odontoid hypoplasia:

- 1 yes
2 no
3 unknown

10. Date of test:

/ /
Month Day Year

Recipient NMDP ID: - -

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11. Was a Mental Development test done pre-transplant?

- 1 yes
2 no
3 unknown

12. Indicate test instrument; report results of test done closest to transplant; report score, not percentile:

- 1 Bayley Scales of Infant Development
2 Stanford Binet Intelligence Scale 4th ed
3 Wechsler Preschool and Primary Scale of Intelligence (WPPSI – Revised)
4 Wechsler Intelligence Scale for Children – III (WISC – III)
5 Other, specify: _____

13. Date of test:

/ /

Month Day Year

14. Full scale score:

15. Verbal score:

16. Performance score:

17. Were the Vineland Adaptive Behavior Scales done pre-transplant?

- 1 yes
2 no
3 unknown

18. Score results:

a. Communication skills:

b. Daily living skills:

c. Sociolizic skills:

19. Date of test:

/ /

Month Day Year

20. Was an eye exam done pre-transplant?

- 1 yes
2 no
3 unknown

21. Visual acuity:

a. Right eye:

/

b. Left eye:

/

22. Was corneal clouding present?

- 1 yes
2 no
3 unknown

23. Date of most recent test:

/ /

Month Day Year

Recipient NMDP ID: - -

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24. Was an audiologic evaluation (auditory brain stem or conditioned response) done pre-transplant?

- 1 yes →
2 no
3 unknown

25. Tympanometry results:

	Normal	Retracted	Flat
a. Right ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>
b. Left ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>

26. Was the hearing loss (HL) in decibels (dB) assessed at the speech threshold for 500 hertz (HZ)?

- 1 yes →
2 no
3 unknown

27. Speech Threshold results at 500 HZ:

	Normal - Mild	Moderate - Moderately Severe	Severe - Profound
a. Right ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>
b. Left ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>

See Degree of Hearing Loss chart below for scale ranges.

28. Was the hearing loss (HL) in decibels (dB) assessed at the speech threshold for 2000 hertz (HZ)?

- 1 yes →
2 no
3 unknown

29. Speech Threshold results at 2000 HZ:

	Normal - Mild	Moderate - Moderately Severe	Severe - Profound
a. Right ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>
b. Left ear:	1 <input type="checkbox"/>	2 <input type="checkbox"/>	3 <input type="checkbox"/>

See Degree of Hearing Loss chart below for scale ranges.

Degree of Hearing Loss: Pure tones and speech testing

Normal:	0-20 dB HL	Moderately Severe:	60-70 dB HL
Mild:	25-40 dB HL	Severe:	75-90 dB HL
Moderate:	45-55 dB HL	Profound:	> 90 dB HL

30. Was pulmonary function testing done pre-transplant?

- 1 yes →
2 no
3 unknown

31. Oxygen saturation on room air: %

32. Results of most recent pulmonary function test:

(If possible, attach a copy of the report.)

- 1 normal
2 abnormal
3 not done

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33. Was an echocardiogram done pre-transplant?

- 1 yes →
- 2 no
- 3 unknown

34. Valvular insufficiency:

- a. Tricuspid:
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
- b. Mitral:
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
- c. Aortic:
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
- d. Pulmonary:
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement

35. Date of test: / /
Month Day Year

36. Was the cardiac contractility tested pre-transplant?

- 1 yes →
- 2 no

37. Ejection fraction: %

38. Shortening fraction: %