In one aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a lysosomal storage disorder (LSD) or one or more symptoms associated with a LSD in a subject in need thereof, wherein the LSD is not Niemann-Pick Type C.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a LSD in a subject in need thereof, wherein the subject is asymptomatic.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for delaying onset of a LSD or one or more symptoms of a LSD that would otherwise be expected to manifest according to typical disease progression.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a LSD or one or more symptoms associated with a LSD in a subject in need thereof, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In a further aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for delaying progression of a LSD or one or more symptoms associated with a LSD over time as compared to typical disease progression, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for reversing progression of a LSD or one or more symptoms associated with a LSD over time, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for improving in a subject

in need thereof a biochemical marker of a LSD over time, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In a further aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for reducing the severity of a LSD or reducing the severity of or eliminating one or more existing symptoms associated with a LSD in a subject in need thereof, wherein the LSD is not Niemann-Pick Type C.

In another aspect, there is disclosed the use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for providing neuroprotection in a subject having, suspected of having, or at risk of having a LSD, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In one embodiment, there is disclosed acetyl-leucine, or a pharmaceutically acceptable salt thereof, for use in a method of treating a LSD or one or more symptoms associated with a LSD in a subject in need thereof, wherein the LSD is not Niemann-Pick Type C.

In one embodiment of the present disclosure, acetyl-leucine, or a pharmaceutically acceptable salt thereof, is disclosed for use in a method of treating a LSD in a subject in need thereof, wherein the subject is asymptomatic.

In another embodiment, there is disclosed acetyl-leucine, or a pharmaceutically acceptable salt thereof, for use in a method of delaying onset of a LSD or one or more symptoms of a LSD that would otherwise be expected to manifest according to typical disease progression.

In a further embodiment, the present disclosure includes acetyl-leucine, or a pharmaceutically acceptable salt thereof, for use in a method of treating a LSD or one or more symptoms associated with a LSD in a subject in need thereof, wherein the method comprises administering a therapeutically effective amount of the acetyl-leucine to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In one embodiment, the present disclosure describes acetyl-leucine, or a pharmaceutically acceptable salt thereof, for use in a method of delaying progression of a LSD or one or more symptoms associated with a LSD over time as compared to typical disease progression, wherein the method comprises administering a therapeutically effective amount of the acetyl-leucine to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In a further embodiment, acetyl-leucine, or a pharmaceutically acceptable salt thereof, is disclosed for use in a method of reversing progression of a LSD or one or more symptoms associated with a LSD over time, wherein the method comprises administering a therapeutically effective amount of the acetyl-leucine to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

In another embodiment, acetyl-leucine, or a pharmaceutically acceptable salt thereof, is disclosed for use in a method of improving in a subject in need thereof a biochemical

Claims:

1. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a lysosomal storage disorder (LSD) or one or more symptoms associated with a LSD in a subject in need thereof, wherein the LSD is not Niemann-Pick Type C.

2. The use according to claim 1, wherein the LSD is chosen from glycogen storage disease, mucopolysaccaridoses, mucolipidoses, oligosaccharidoses, lipidoses, sphingolipidoses, and lysosomal transport diseases.

3. The use according to claim 1, wherein the LSD is chosen from a primary lysosomal hydrolase defect, a post-translational processing defect of lysosomal enzymes, a trafficking defect for lysosomal enzymes, a defect in lysosomal enzyme protection, a defect in soluble non-enzymatic lysosomal proteins, a transmembrane (non-enzyme) protein defect, and an unclassified defect.

4. The use according to claim 1, wherein the LSD is chosen from Tay-Sachs disease, the AB variant of Tay-Sachs disease, Sandhoff disease, Niemann-Pick type A disease, Niemann-Pick type B disease, Fabry disease, neuronal ceroid lipofuscinoses, Krabbe disease, Farber disease, Gaucher disease, metachromatic leukodystrophy, multiple sulphatase deficiency, mucolipidosis II, mucolipidosis III, MPS III, MPS VII, GM1 gangliosidosis, and aspartylglucosaminuria.

5. The use according to claim 1, wherein the LSD is chosen from Tay-Sachs disease, the AB variant of Tay-Sachs disease, Sandhoff disease, Niemann Pick type A disease, mucolipidosis II, mucolipidosis III, MPS III, MPS VII, GM1 gangliosidosis, and aspartylglucosaminuria.

6. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a LSD in a subject in need thereof, wherein the subject is asymptomatic.

7. The use according to claim 6, wherein the subject has been found to have a genetic and/or biochemical marker of the LSD.

8. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for delaying onset of a LSD or one or more symptoms of a LSD that would otherwise be expected to manifest according to typical disease progression.

9. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for treating a LSD or one or more symptoms associated with a LSD in a subject in need thereof, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

10. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for delaying progression of a LSD or one or more symptoms associated with a LSD over time as compared to typical disease progression, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

11. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for reversing progression of a LSD or one or more symptoms associated with a LSD over time, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

12. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for improving in a subject in need thereof a biochemical marker of a LSD over time, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

13. The use according to claim 12, wherein the biochemical marker is increased lysosomal volume.

14. The use according to any of claims 1-13, wherein a therapeutically effective amount of acetyl-leucine is to be administered to the subject in need thereof when the subject is asymptomatic.

15. The use according to claim 14, wherein the therapeutically effective amount of acetyl-leucine is to be administered after the subject has been found to have a genetic and/or biochemical marker of the LSD.

16. The use according to any of claims 1-8, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject in need thereof for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.

17. The use according to any of claims 1-16, wherein the acetyl-leucine is acetyl-DL-leucine.

18. The use according to any of claims 1-16, wherein the acetyl-leucine has an enantiomeric excess of the L-enantiomer or the D-enantiomer.

19. The use according to any of claims 1-16, wherein the acetyl-leucine is in a single enantiomeric form of either the L-enantiomer or the D-enantiomer.

20. The use according claim 19, wherein the single enantiomeric form is the L-enantiomer.

21. The use according to any of claims 1-8, wherein the acetyl-leucine is to be administered to the subject in need thereof at a therapeutically effect amount of from about 1 g to about 15 g per day, from about 1 g to about 10 g per day, from about 1.5 g to about 7 g per day, from about 4 g to about 6 g per day, or from about 4 g to about 5 g per day.

22. The use according to any of claims 9-16, wherein the therapeutically effective amount of acetyl-leucine is from about 1 g to about 15 g per day, from about 1 g to about 10 g per day, from about 1.5 g to about 7 g per day, from about 4 g to about 6 g per day, or from about 4 g to about 5 g per day.

23. The use according to any of claims 6-13, wherein the LSD is chosen from glycogen storage disease, mucopolysaccaridoses, mucolipidoses, oligosaccharidoses, lipidoses, sphingolipidoses, and lysosomal transport diseases.

24. The use according to any of claims 6-13, wherein the LSD is chosen from a primary lysosomal hydrolase defect, a post-translational processing defect of lysosomal enzymes, a trafficking defect for lysosomal enzymes, a defect in lysosomal enzyme protection, a defect in soluble non-enzymatic lysosomal proteins, a transmembrane (non-enzyme) protein defect, and an unclassified defect.

25. The use according to any of claims 6-13, wherein the LSD is chosen from Niemann-Pick type C disease, Tay-Sachs disease, the AB variant of Tay-Sachs disease, Sandhoff disease, Niemann-Pick type A disease, Niemann-Pick type B disease, Fabry disease, neuronal ceroid lipofuscinoses, Krabbe disease, Farber disease, Gaucher disease, metachromatic leukodystrophy, multiple sulphatase deficiency, mucolipidosis II, mucolipidosis III, MPS III, MPS VII, GM1 gangliosidosis, and aspartylglucosaminuria

26. The use according to any of claim 6-13, wherein the LSD is chosen from Niemann-Pick type C disease, Tay-Sachs disease, the AB variant of Tay-Sachs disease, Sandhoff disease, Niemann Pick type A disease, mucolipidosis II, mucolipidosis III, MPS III, MPS VII, GM1 gangliosidosis, and aspartylglucosaminuria

27. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for reducing the severity of a LSD or reducing the severity of or eliminating one or more existing symptoms associated with a LSD in a subject in need thereof, wherein the LSD is not Niemann-Pick Type C.

28. Use of acetyl-leucine, or a pharmaceutically acceptable salt thereof, in the manufacture of a medicament, for providing neuroprotection in a subject having, suspected of having, or at risk of having a LSD, wherein a therapeutically effective amount of the acetyl-leucine is to be administered to the subject for a duration chosen from at least about 3 months, at least about 6 months, at least about 1 year, at least about 2 years, and at least about 5 years.