

CLAIMS

1. A method of screening for a fetal trisomy in a pregnant female subject, the method comprising:
 - detecting one or more lipid markers in a sample obtained from the subject in the first trimester of pregnancy, wherein the one or more lipid markers is indicative of the risk of a fetal trisomy in the subject; and
 - determining the risk of a fetal trisomy in the pregnant female subject on the basis of the one or more lipid markers detected.
2. The method according to claim 1, wherein the one or more markers is indicative of an increased risk of a fetal trisomy, a low or normal risk of a fetal trisomy, and/or the presence or absence of a fetal trisomy.
3. The method according to claim 1 or 2, wherein the fetal trisomy comprises trisomy 21.
4. The method according to any one of claims 1 to 3, wherein the one or more lipid markers comprise one or more of a phospholipid, a glycerolipid, a glycerophospholipid, a sphingolipid, a ceramide, a sterol, a glycosphingolipid, a dolicol, a lysolipid, a fatty acid, a triacylglyceride, a diacylglycerides, a monacylglycerides, an isoprenoid, a prostanoid, an eicosanoid, a sterol derivatives, a prenol lipid, a saccharolipid, a saturated fatty acid, a long chain saturated fatty acid, an unsaturated fatty acid, a polyunsaturated fatty acid, a long chain polyunsaturated fatty acid, a cholesterol and isomers thereof, a dehydrocholesterol and isomers thereof, a cholesterol esters and isomers thereof, a dehydrocholesterol esters and isomers thereof, a lyso derivative of any of the aforementioned lipid markers, and/or a fragment of any of the aforementioned lipid markers.
5. The method according to any one of claims 1 to 4, wherein the method comprises detecting the one or more lipid markers by mass spectrometry.

6. The method according to claim 5, wherein the one or more lipid markers comprise a lipid with one or more ions with the following assigned m/z : 703.5580, 369.3491, 367.3312, 680.5953, 687.5651, 445.3648, 445.3633, 727.5567, 429.3706, 1202.3250, 431.3875, 429.3726, 844.6007, 688.5950, 890.7703, 1276.3440, 711.5619, 1128.3140, 670.6080, and/or one or more of the aforementioned lipid markers with a mass tolerance of ± 20 ppm, and/or a similar m/z and/or retention time.

7. The method according to claim 5, wherein the one or more lipid markers comprise a lipid with one or more ions with the following assigned m/z : 340.3576, 355.2511, 363.2531, 390.7783, 399.3453, 404.7929, 407.2269, 408.3136, 427.3771, 429.2121, 447.3448, 447.3460, 461.3620, 469.3597, 471.4063, 473.3184, 478.3545, 481.3239, 489.4134, 506.4412, 530.5135, 544.5071, 551.5172, 555.4646, 558.5448, 568.4784, 579.4305, 603.4949, 604.5660, 607.5640, 618.5444, 626.4976, 640.5345, 640.5699, 647.5577, 676.4879, 684.5594, 700.5542, 728.5875, 740.5176, 744.5807, 772.6135, 780.5458, 788.6118, 798.5637, 817.5510, 816.6401, 820.5947, 826.5947, 832.6303, 834.5978, 850.5523, 860.6632, 864.5363, 876.6591, 902.5855, 904.6889, 904.6932, 920.6888, 928.5873, 948.7143, 964.7115, 992.7408, 1008.7339, 1036.7696, 1052.7680, 1078.9686, 670.6080, and/or one or more of the aforementioned lipid markers with a mass tolerance of ± 20 ppm, and/or a similar m/z .

8. The method according to any one of claims 1 to 7, wherein the sample comprises one or more of blood, plasma and serum.

9. The method according to any one of claims 1 to 8, wherein the one or more lipid markers comprise at least two lipid markers.

10. The method according to any one of claims 1 to 9, wherein the one or more lipid markers comprise a combination of lipid markers.

11. The method according to any one of claims 1 to 10, wherein detecting one or more lipid markers comprises detecting the level of the one or more lipid markers.

12. The method according to any one of claims 1 to 11, wherein detecting one or more lipid markers further comprises comparing the one or more lipid markers with one or more control lipid markers from a sample obtained from a subject in the first trimester of a pregnancy without a fetal trisomy and/or with one or more control lipid markers from a sample obtained from a subject in the first trimester of a pregnancy with a fetal trisomy.

13. The method according to any one of claims 1 to 12, wherein the method comprises using a computer processor means to process data associated with the presence and/or level of the one or more lipid markers to generate a likelihood and/or risk of the presence or absence of a fetal trisomy.

14. The method according to claim 13, wherein the method comprises transferring the data over the internet to the computer processing means.

15. The method according to any one of claims 1 to 14, wherein the method is used to identify the presence or absence of a fetal trisomy, to identify a fetal trisomy in the subject, to exclude the presence of a fetal trisomy in the subject, to identify an increased risk of a fetal trisomy, and/or to identify a low or normal risk of a fetal trisomy.

16. A computer-readable medium encoded with programming instructions executable by a computer processor means to allow the computer processor means to receive data associated with the presence and/or level of one or more lipid markers in a sample obtained from a subject in the first trimester of pregnancy and indicative of the risk of a fetal trisomy in a subject and process the data to generate a likelihood and/or risk of the presence or absence of a fetal trisomy in the subject.

17. A computer processor means comprising a computer-readable medium according to claim 16.

18. A system when used to determine the risk of a fetal trisomy in a subject, the system comprising a computer processor having a computer-readable medium encoded with programming instructions executable by the computer processor means to allow the computer processor means to receive data associated with the presence and/or level of one

or more lipid markers in a sample obtained from a subject in the first trimester of pregnancy and indicative of the risk of a fetal trisomy in a subject and process the data to generate a likelihood and/or risk of the fetal trisomy in the subject.

19. The system according to claim 18, wherein the data is transferred over the internet to the computer processing means.

20. The system according to claim 18 or 19, wherein the system comprises a mass spectrograph.

21. A method of determining the risk of a pregnant female subject carrying a fetus with a trisomy, the method comprising:

detecting one or more lipid markers in a sample obtained from the subject in the first trimester of pregnancy, wherein the one or more markers is indicative of the risk of a fetal trisomy in the subject; and

determining the risk of the subject carrying a fetus with a trisomy on the basis of the one or more lipid markers detected.

22. A method according to claim 21, wherein the determining of the risk comprises a computer processor means to process data associated with the presence and/or level of the one or more lipid markers.

23. A kit when used to perform the method of any one of claims 1 to 15, 21 or 22.

24. A method of identifying one or more lipid markers indicative of the risk of a fetal trisomy, the method comprising:

identifying one or more lipid markers which are differentially present between a sample obtained in the first trimester of pregnancy from a pregnant female subject having a fetal trisomy and a sample obtained in the first trimester of pregnancy from a pregnant female subject without a fetal trisomy; and

identifying the one or more lipid markers as one or more lipid markers indicative of the risk of a fetal trisomy.

25. A method of identifying one or more lipid markers indicative of the risk of a fetal trisomy, the method comprising identifying a plurality of lipid markers in a sample obtained in the first trimester of pregnancy which in combination are indicative of the risk of a fetal trisomy.