

CLAIMS

1. A method of screening for a fetal trisomy 21 in a pregnant female subject, the method comprising:

detecting a specific lipid marker by mass spectrometry in a sample containing serum obtained from the subject in the first trimester of pregnancy;

comparing the presence and/or level of the specific lipid marker with the presence and/or level of a lipid indicative of the risk of fetal trisomy 21; and

determining the risk of a fetal trisomy 21 in the pregnant female subject on the basis of the comparison of the presence and/or level of the specific lipid marker with the presence and/or level of the lipid indicative of the risk of a fetal trisomy 21;

wherein the lipid indicative of the risk of the fetal trisomy 21 comprises: a lipid indicative of an increased risk of a fetal trisomy 21; and/or a lipid indicative of a decreased risk of a fetal trisomy 21; and/or a lipid indicative of the presence of a fetus with a fetal trisomy 21 in a pregnant female subject; and/or a lipid indicative of a fetus without a fetal trisomy 21 in a pregnant female subject.

2. The method according to claim 1, wherein the specific lipid marker is indicative of an increased risk of a fetal trisomy 21, a low or normal risk of a fetal trisomy 21, and/or the presence or absence of a fetal trisomy 21.

3. The method according to claims 1 or 2, wherein the specific lipid marker comprises 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.

4. The method according to any one of claims 1 to 3, wherein the specific lipid marker comprises a lipid with an ion 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 of the aforementioned lipid markers with a mass tolerance of ± 20 ppm, and/or a similar m/z .

5. The method according to any one of claims 1 to 3, wherein the specific lipid marker comprises a lipid with an ion 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 of the aforementioned lipid markers with a mass tolerance of ± 20 ppm, and/or a similar m/z .

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6. The method according to any one of claims 1 to 5, wherein the specific lipid marker comprises at least two specific lipid markers.

7. The method according to any one of claims 1 to 6, wherein the specific lipid marker comprises a combination of specific lipid markers.

8. The method according to any one of claims 1 to 7, wherein the method comprises using a computer processor means to process data associated with the presence and/or level of the specific lipid marker to generate a likelihood and/or risk of the presence or absence of a fetal trisomy 21.

9. The method according to claim 8, wherein the method comprises transferring the data over the internet to the computer processing means.

10. The method according to any one of claims 1 to 9, wherein the method is used to identify the presence or absence of a fetal trisomy 21, to identify an increased risk of a fetal trisomy 21, and/or to identify a low or normal risk of a fetal trisomy 21.

11. 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 a specific lipid marker in a sample containing serum obtained from a subject in the first trimester of pregnancy, detected by mass spectrometry and indicative of the risk of a fetal trisomy 21 in a subject and process the data to generate a likelihood and/or risk of the presence or absence of a fetal trisomy 21 in the subject.

12. A computer processor means comprising a computer-readable medium according to claim 11.

13. A system when used to determine the risk of a fetal trisomy 21 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 a specific lipid marker in a sample containing serum obtained from a subject in the first trimester of pregnancy, detected by mass spectrometry and indicative of the risk of a fetal trisomy 21 in a subject and process the data to generate a likelihood and/or risk of the fetal trisomy 21 in the subject.

14. The system according to claim 13, wherein the data is transferred over the internet to the computer processing means.

15. The system according to claim 13 or 14, wherein the system comprises a mass spectrograph.

16. A method of determining the risk of a pregnant female subject carrying a fetus with a trisomy 21, the method comprising:
detecting a specific lipid marker by mass spectrometry in a sample containing

serum obtained from the subject in the first trimester of pregnancy, wherein the specific lipid marker is indicative of the risk of a fetal trisomy 21 in the subject; and
determining the risk of the subject carrying a fetus with a trisomy 21 on the basis of the specific lipid marker detected.

17. A method according to claim 16, wherein the determining of the risk comprises a computer processor means to process data associated with the presence and/or level of the specific lipid marker.

18. A kit when used to perform the method of any one of claims 1 to 10, 16 or 17, wherein the kit comprises an isotopically labelled lipid.

19. A method of identifying a lipid marker indicative of the risk of a fetal trisomy 21, the method comprising:

identifying a specific lipid marker detected by mass spectrometry which is differentially present between a sample containing serum obtained in the first trimester of pregnancy from a pregnant female subject having a fetal trisomy 21 and a sample containing serum obtained in the first trimester of pregnancy from a pregnant female subject without a fetal trisomy 21; and
identifying the specific lipid marker as a lipid marker indicative of the risk of a fetal trisomy 21.

20. A method of identifying a combination of lipid markers in a sample containing serum indicative of the risk of a fetal trisomy 21, the method comprising identifying a plurality of specific lipid markers in the sample containing serum obtained in the first trimester of pregnancy, detected by mass spectrometry and which in combination are indicative of the risk of a fetal trisomy 21.