CLAIMS

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

detecting one or more specific lipid markers by mass spectrometry in a sample obtained from the subject in the first trimester of pregnancy;

comparing the presence and/or level of the one or more specific lipid markers with the presence and/or level of one or more lipids known to be 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 one or more specific lipid markers with the presence and/or level of the one or more lipids known to be indicative of the risk of a fetal trisomy 21.

2. The method according to claim 1, wherein the one or more specific lipid markers 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 one or more specific lipid markers comprise one or more of a phoshopholipid, 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 one or more specific 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*.

5. The method according to any one of claims 1 to 3, wherein the one or more specific 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*.

6. The method according to any one of claims 1 to 5, wherein the sample comprises one or more of blood, plasma and serum.

7. The method according to any one of claims 1 to 6, wherein the one or more specific lipid markers comprise at least two specific lipid markers.

8. The method according to any one of claims 1 to 7, wherein the one or more specific lipid markers comprise a combination of specific lipid markers.

9. The method according to any one of claims 1 to 8, wherein the detecting of the one or more lipid specific markers comprises detecting the level of the one or more specific lipid markers.

10. The method according to any one of claims 1 to 9, wherein the method comprises using a computer processor means to process data associated with the presence and/or level of the one or more specific lipid markers to generate a likelihood and/or risk of the presence or absence of a fetal trisomy 21.

11. The method according to claim 10, wherein the method comprises transferring the data over the internet to the computer processing means.

12. The method according to any one of claims 1 to 11, 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.

13. 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 specific lipid markers in a sample 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.

14. A computer processor means comprising a computer-readable medium according to claim 13.

15. 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 one or more specific lipid markers in a sample 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.

16. The system according to claim 15, wherein the data is transferred over the internet to the computer processing means.

17. The system according to claim 15 or 16, wherein the system comprises a mass spectrograph.

18. A method of determining the risk of a pregnant female subject carrying a fetus

with a trisomy 21, the method comprising:

detecting one or more specific lipid markers in a sample obtained from the subject in the first trimester of pregnancy, wherein the one or more specific lipid markers 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 one or more specific lipid markers detected.

19. A method according to claim 18, 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 specific lipid markers.

20. A kit when used to perform the method of any one of claims 1 to 12, 18 or 19.

21. A method of identifying one or more lipid markers indicative of the risk of a fetal trisomy 21, the method comprising:

identifying one or more specific lipid markers detected by mass spectrometry which are differentially present between a sample obtained in the first trimester of pregnancy from a pregnant female subject having a fetal trisomy 21 and a sample obtained in the first trimester of pregnancy from a pregnant female subject without a fetal trisomy 21; and

identifying the one or more specific lipid markers as one or more lipid markers indicative of the risk of a fetal trisomy 21.

22. A method of identifying one or more lipid markers indicative of the risk of a fetal trisomy 21, the method comprising identifying a plurality of specific lipid markers in a sample 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.