

What is claimed:

1. A method for determining pattern of inheritance in an embryo for a region of interest, comprising:
 - receiving embryo, maternal and paternal sequence data, wherein the maternal sequence data is from a mother of the embryo and the paternal sequence data is from a father of the embryo;
 - aligning the received sequence data to a reference genome;
 - identifying a region of interest in the aligned embryo sequence data;
 - identifying single nucleotide polymorphisms (SNPs) in the maternal sequence data, paternal sequence data, and the identified region of interest in the embryo sequence data;
 - imputing missing gaps in the maternal sequence data and the paternal sequence data using an imputation reference;
 - counting the number of SNPs that are common between the embryo and the mother in the identified region of interest for the embryo sequence data and a corresponding region on the maternal sequence data to determine a maternal contribution value;
 - counting the number of SNPs that are common between the embryo and the father in the identified region of interest for the embryo sequence data and a corresponding region on the paternal sequence data to determine a paternal contribution value; and
 - classifying a pattern of inheritance for the embryo as maternal or paternal based on the relative contribution values between mother and father.
2. The method of Claim 1, further comprising counting SNPs across the entire genome to determine the maternal and paternal contribution values and determine if the embryo is polyploid.
3. The method of Claim 2, wherein for an embryo that is polyploid, the method further comprising classifying a pattern of inheritance for the polyploid as maternal or paternal based on the relative contribution values between mother and father.

4. The method of Claim 1, wherein at least one of the embryo, maternal and paternal sequence data is acquired by low-coverage sequencing.
5. The method of Claim 4, wherein the low-coverage sequencing is between about 0.001 and 10x.
6. The method of Claim 4, wherein the low-coverage sequencing is between about 0.01 and 0.5x.
7. The method of Claim 4, wherein the low-coverage sequencing is between about 0.25 and 0.2x.
8. The method of Claim 1, wherein the region of interest is genome wide.
9. The method of Claim 1, wherein the region of interest is a copy number variation.
10. The method of Claim 1, wherein the imputation reference comprises at least 1000 genomes.
11. The method of Claim 1, further comprising filtering at least one of the embryo, maternal and paternal sequencing data to remove sequencing artifacts.
12. The method of Claim 11, wherein the filtering comprises excluding sequences of sites with any missing alleles between the embryo, mother and father.
13. The method of Claim 11, wherein the filtering comprises excluding sequences of sites with constant alleles across between the embryo, mother and father.
14. The method of Claim 11, wherein the filtering comprises excluding sequences of sites with a novel allele within one of the embryo, mother and father.
15. The method of Claim 11, wherein the filtering comprises excluding SNPs that are inconsistent with Mendelian inheritance.
16. A non-transitory computer-readable medium storing computer instructions for determining pattern of inheritance in an embryo for a region of interest, comprising:

receiving embryo, maternal and paternal sequence data, wherein the maternal sequence data is from a mother of the embryo and the paternal sequence data is from a father of the embryo;

aligning the received sequence data to a reference genome;

identifying a region of interest in the aligned embryo sequence data;

identifying single nucleotide polymorphisms (SNPs) in the maternal sequence data, paternal sequence data, and the identified region of interest in the embryo sequence data;

imputing missing gaps in the maternal sequence data and the paternal sequence data using an imputation reference;

counting the number of SNPs that are common between the embryo and the mother in the identified region of interest for the embryo sequence data and a corresponding region on the maternal sequence data to determine a maternal contribution value;

counting the number of SNPs that are common between the embryo and the father in the identified region of interest for the embryo sequence data and a corresponding region on the paternal sequence data to determine a paternal contribution value; and

classifying a pattern of inheritance for the embryo as maternal or paternal based on the relative contribution values between mother and father.

17. The method of Claim 16, further comprising counting SNPs across the entire genome to determine the maternal and paternal contribution values and determine if the embryo is polyploid.
18. The method of Claim 17, wherein for an embryo that is polyploid, the method further comprising classifying a pattern of inheritance for the polyploid as maternal or paternal based on the relative contribution values between mother and father.
19. The method of Claim 16, wherein at least one of the embryo, maternal and paternal sequence data is acquired by low-coverage sequencing.
20. The method of Claim 19, wherein the low-coverage sequencing is between about 0.001 and 10x.
21. The method of Claim 19, wherein the low-coverage sequencing is between about 0.01 and 0.5x.
22. The method of Claim 19, wherein the low-coverage sequencing is between about 0.25 and 0.2x.
23. The method of Claim 16, wherein the region of interest is genome wide.

24. The method of Claim 16, wherein the region of interest is a copy number variation.
25. The method of Claim 16, wherein the imputation reference comprises at least 1000 genomes.
26. The method of Claim 16, further comprising filtering at least one of the embryo, maternal and paternal sequencing data to remove sequencing artifacts.
27. The method of Claim 26, wherein the filtering comprises excluding sequences of sites with any missing alleles between the embryo, mother and father.
28. The method of Claim 26, wherein the filtering comprises excluding sequences of sites with constant alleles across between the embryo, mother and father.
29. The method of Claim 26, wherein the filtering comprises excluding sequences of sites with a novel allele within one of the embryo, mother and father.
30. The method of Claim 26, wherein the filtering comprises excluding SNPs that are inconsistent with Mendelian inheritance.
31. A system for determining pattern of inheritance in an embryo for a region of interest, comprising:

a data store for receiving embryo, maternal and paternal sequence data, wherein the maternal sequence data is from a mother of the embryo and the paternal sequence data is from a father of the embryo;

a computing device communicatively connected to the data store, the computing device comprising

an ROI engine configured to align the received sequence data to a reference genome, and identify a region of interest in the aligned embryo sequence data;

a SNP identification engine configured to identify single nucleotide polymorphisms (SNPs) in the maternal sequence data, paternal sequence data, and the identified region of interest in the embryo sequence data;

an imputation engine configured to impute missing gaps in the maternal sequence data and the paternal sequence data using an imputation reference; and

a POI engine configured to:

count the number of SNPs that are common between the embryo and the mother in the identified region of interest for the embryo sequence data and a corresponding region on the maternal sequence data to determine a maternal contribution value;

count the number of SNPs that are common between the embryo and the father in the identified region of interest for the embryo sequence data and a corresponding region on the paternal sequence data to determine a paternal contribution value; and

classify a pattern of inheritance for the embryo as maternal or paternal based on the relative contribution values between mother and father;

and

a display communicatively connected to the computing device and configured to display a report containing the classified pattern of inheritance for the embryo.

32. The system of Claim 31, wherein the POI engine is further configured to count SNPs across the entire genome to determine the maternal and paternal contribution values and determine if the embryo is polyploid.
33. The system of Claim 32, wherein for an embryo that is polyploid, the POI engine is further configured to classify a pattern of inheritance for the polyploid as maternal or paternal based on the relative contribution values between mother and father.
34. The system of Claim 31, wherein at least one of the embryo, maternal and paternal sequence data is acquired by low-coverage sequencing.
35. The system of Claim 34, wherein the low-coverage sequencing is between about 0.001 and 10x.
36. The system of Claim 34, wherein the low-coverage sequencing is between about 0.01 and 0.5x.
37. The system of Claim 34, wherein the low-coverage sequencing is between about 0.25 and 0.2x.

38. The system of Claim 31, wherein the region of interest is genome wide.
39. The system of Claim 31, wherein the region of interest is a copy number variation.
40. The system of Claim 31, wherein the imputation reference comprises at least 1000 genomes.
41. The system of Claim 31, wherein the imputation engine is further configured to filter at least one of the embryo, maternal and paternal sequencing data to remove sequencing artifacts.
42. The system of Claim 41, wherein the filtering comprises excluding sequences of sites with any missing alleles between the embryo, mother and father.
43. The system of Claim 41, wherein the filtering comprises excluding sequences of sites with constant alleles across between the embryo, mother and father.
44. The system of Claim 41, wherein the filtering comprises excluding sequences of sites with a novel allele within one of the embryo, mother and father.
45. The system of Claim 41, wherein the filtering comprises excluding SNPs that are inconsistent with Mendelian inheritance.