

Trio解析

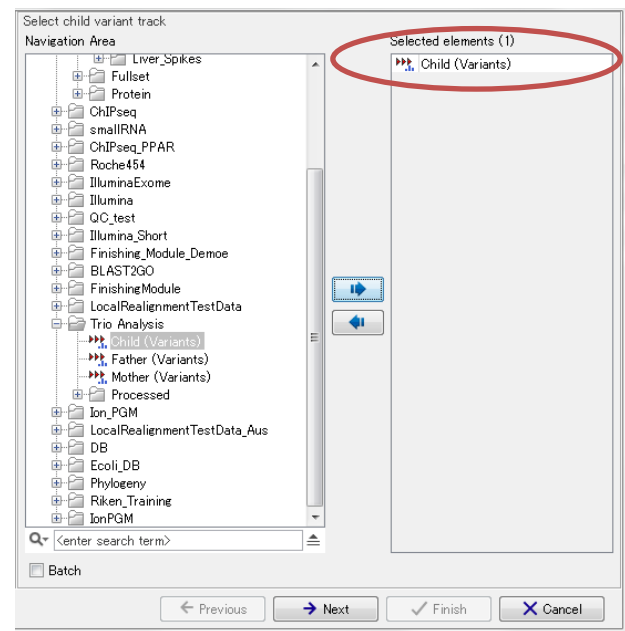
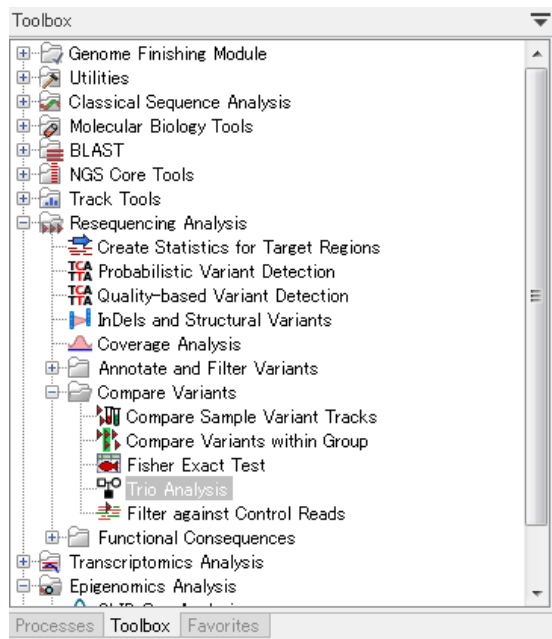
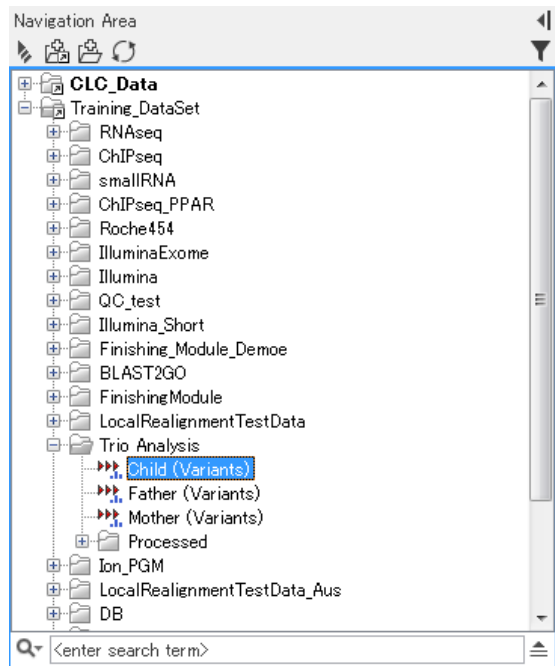


Trio 解析

- Trio 解析では、両親と病気のお子様といった家族間での変異が新規のものなのか(de novo), 両親どちらかから受け継いだものなのか (Inherited from Mother, Inherited from Father), 両親はヘテロだけでも、子供にはホモとして現れた(Accumulated)といった変異の状態をみつけるために使用します。
- Trio解析をご利用する前に、まず母親、父親、子供それぞれの変異の検出が行われている必要があります。



Trio 解析



- Navigation Areaから子供の変異データを選択。
- Toolboxから Resequencing Analysis > Compare Variants > Trio Analysis を選択、ダブルクリック。
- ウィザードが起動し、選択したデータが選ばれていることを確認。



Trio 解析

The screenshot shows the 'Trio Analysis' window at step 2. The left sidebar lists '1. Select child variant track' and '2. Trio analysis parameters'. The main area is titled 'Trio analysis parameters' and contains the following sections:

- Parents:** Two text input fields for 'Mother (Variants)' and 'Father (Variants)', each with a file selection icon.
- Full human genome parameters:** A checked checkbox for 'Full human genome'.
- Child gender:** Radio buttons for 'Male' and 'Female', with 'Female' selected.
- Chromosome names:** Three dropdown menus for 'Chromosome X name', 'Chromosome Y name', and 'Chromosome M name', all set to 'Ignore'.

At the bottom, there are navigation buttons: '?', a refresh icon, 'Previous', 'Next', 'Finish', and 'Cancel'.

- 両親の変異を選択
- 全ゲノムデータかどうか
- 子供の性
- X染色体、Y染色体、ミトコンドリアがどのように名前を付けられているか(ゲノムトラックに付けられている名前)

The screenshot shows the 'Trio Analysis' window at step 3. The left sidebar lists '1. Select child variant track', '2. Trio analysis parameters', and '3. Result handling'. The main area is titled 'Result handling' and contains the following sections:

- Result handling:** Radio buttons for 'Open' and 'Save', with 'Save' selected.
- Log handling:** A checked checkbox for 'Open log'.

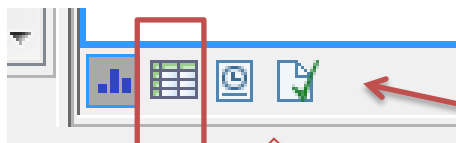
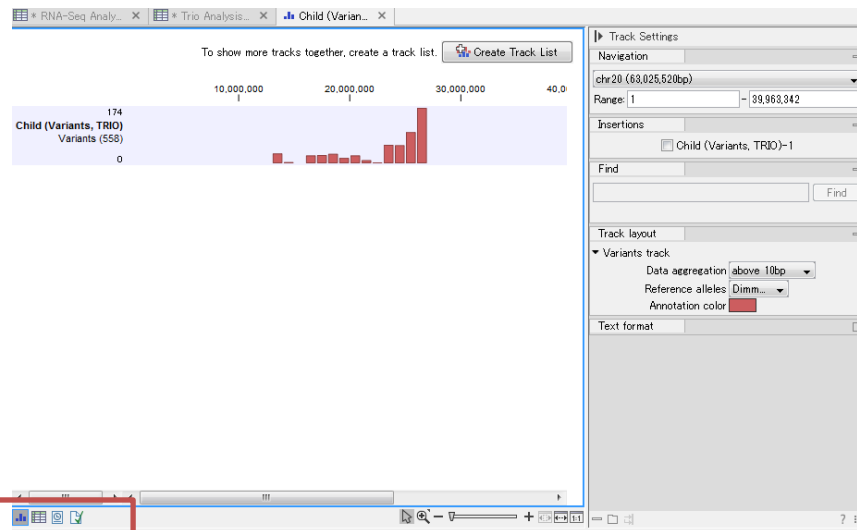
At the bottom, there are navigation buttons: '?', a refresh icon, 'Previous', 'Next', 'Finish', and 'Cancel'.

- データ保存やログについて選択



結果

- Trio Analysis
 - Child (Variants)
 - Father (Variants)
 - Mother (Variants)
 - Child (Variants, TRIO)



Rows: 988 Table view Genome

Forward/rev...	Average qua...	Alteration (Mother)	Alteration (Father)	Zygosity (Mother)	Zygosity (Father)	Inheritance	Mendelian inheritance problem
42	0.44	24.40 A > C	A > C	Homozygous	Homozygous	Inherited from both	No
14	0.45	23.70 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
23	0.44	24.10 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
42	0.26	24.60 C > T	C > T	Homozygous	Homozygous	Inherited from both	No
15	0.38	24.00 C > G	C > G	Homozygous	Homozygous	Inherited from both	No
94	0.49	29.30 T > G	T > G	Homozygous	Homozygous	Inherited from both	No
39	0.43	29.70 A > G	A > G	Homozygous	Homozygous	Inherited from both	No
86	0.18	30.80 G > A	G > A	Homozygous	Homozygous	Inherited from both	No
2	0.29	24.40 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.20	29.80 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
8	0.27	28.00 T > C	T > C	Homozygous	Homozygous	Inherited from both	No
1	0.06	24.20 T > C, T > T	T > C, T > T	Heterozygous	Heterozygous	Accumulative	No
1	0.09	29.00 A > A, A > C	A > A, A > C	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.10	27.90 A > A, A > C	A > A, A > C	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.11	24.60 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Accumulative	No
25	0.07	24.30 A > A, A > T	A > A, A > T	Heterozygous	Heterozygous	Inherited from either parent	No
23	0.08	23.60 A > A, A > T	A > A, A > T	Heterozygous	Heterozygous	Inherited from either parent	No
7	0.42	24.50 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
4	0.43	25.70 C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.07	24.20 T > C	T > C	Homozygous	Homozygous	Inherited from both	No
4	0.29	23.80 A > G	A > G	Homozygous	Homozygous	Inherited from both	No
6	0.14	25.30 T > G, T > T	T > G, T > T	Heterozygous	Heterozygous	Accumulative	No
1	0.12	25.60 A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Accumulative	No
1	0.20	26.20 A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.20	29.80 A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Inherited from either parent	No

Create Track from Selection

Table Settings

- Region
- Type
- Reference
- Allele
- Reference allele
- Length
- Linkage
- Zygosity
- Count
- Coverage
- Frequency
- Probability
- Forward read count
- Reverse read count
- Forward/reverse balance
- Average quality
- Alteration (Mother)
- Alteration (Father)
- Zygosity (Mother)
- Zygosity (Father)
- Inheritance
- Mendelian inheritance problem

Select All

Deselect All

結果: 子供の変異情報の閲覧

Rows: 558 Table view: Genome

...	Forward/rev...	Average qua...	Alteration (Mother)	Alteration (Father)	Zygosity (Mother)	Zygosity (Father)	Inheritance	Mendelian inheritance problem
42	0.44	24.40	A > C	A > C	Homozygous	Homozygous	Inherited from both	No
14	0.45	23.70	C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
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39	0.43	29.70	A > G	A > G	Homozygous	Homozygous	Inherited from both	No
36	0.18	30.80	G > A	G > A	Homozygous	Homozygous	Inherited from both	No
2	0.29	24.40	C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.20	29.80	C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
8	0.27	28.00	T > C	T > C	Homozygous	Homozygous	Inherited from both	No
1	0.06	24.20	T > C, T > T	T > C, T > T	Heterozygous	Heterozygous	Accumulative	No
1	0.09	29.00	A > A, A > C	A > A, A > C	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.10	27.90	A > A, A > C	A > A, A > C	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.11	24.60	C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Accumulative	No
25	0.07	24.30	A > A, A > T	A > A, A > T	Heterozygous	Heterozygous	Inherited from either parent	No
23	0.08	23.60	A > A, A > T	A > A, A > T	Heterozygous	Heterozygous	Inherited from either parent	No
7	0.42	24.50	C > C, C > T	C > C, C > T	Heterozygous	Heterozygous	Inherited from either parent	No
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1	0.07	24.20	T > C	T > C	Homozygous	Homozygous	Inherited from both	No
4	0.29	23.80	A > G	A > G	Homozygous	Homozygous	Inherited from both	No
6	0.14	25.30	T > G, T > T	T > G, T > T	Heterozygous	Heterozygous	Accumulative	No
1	0.12	25.60	A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Accumulative	No
1	0.20	26.20	A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Inherited from either parent	No
1	0.20	29.80	A > A, A > G	A > A, A > G	Heterozygous	Heterozygous	Inherited from either parent	No

Table Settings

- Region
- Type
- Reference
- Allele
- Reference allele
- Length
- Linkage
- Zygosity
- Count
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- Frequency
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- Forward read count
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- Forward/reverse balance
- Average quality
- Alteration (Mother)
- Alteration (Father)
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- Zygosity (Father)
- Inheritance
- Mendelian inheritance problem

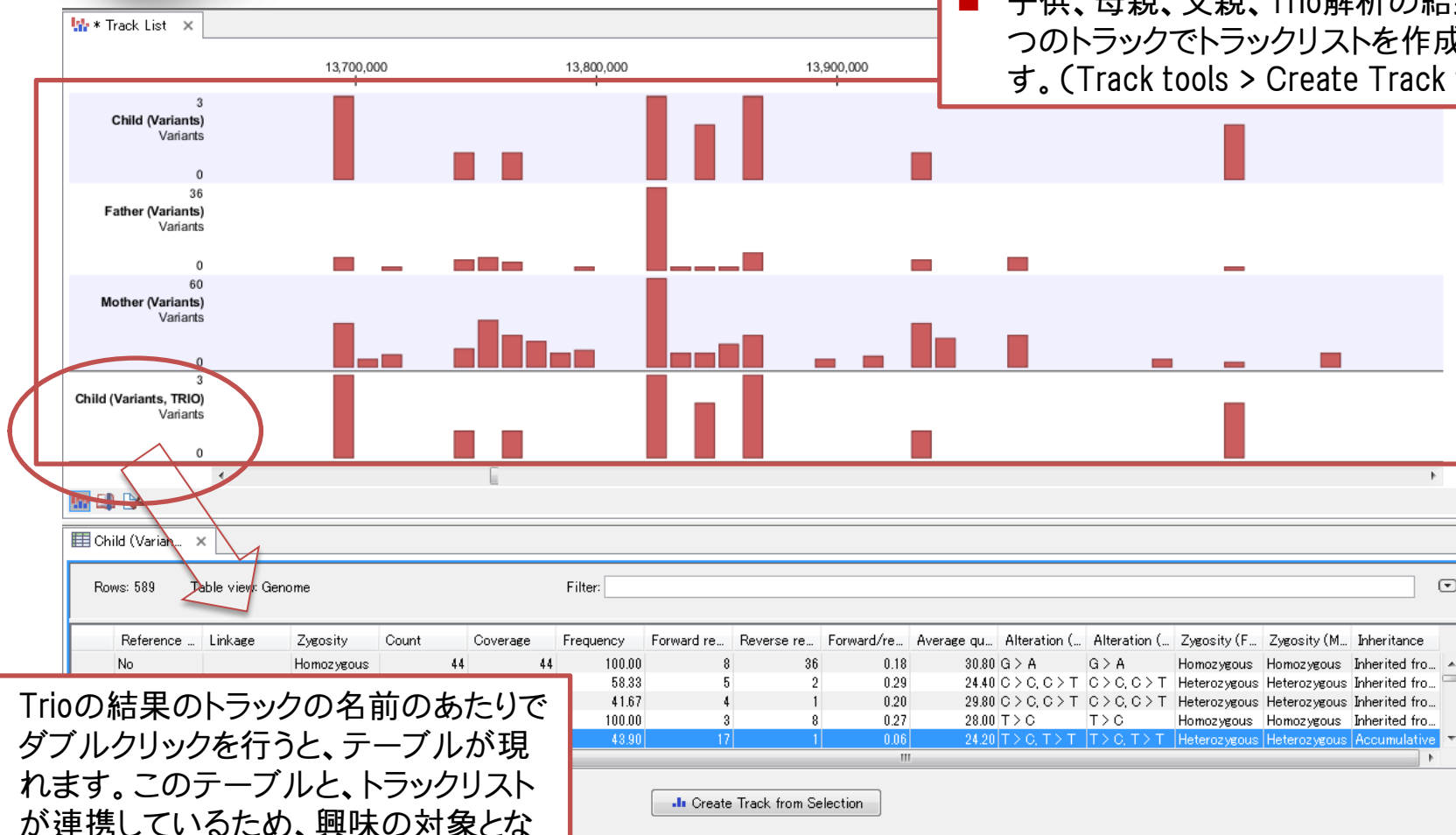
Select All
Deselect All

Create Track from Selection

■ 両親のZygosityや子供の変異がDe Novoなのか、両親由来なのか、といった情報が一番右の列に表示されます。

結果

■ 子供、母親、父親、Trio解析の結果と4つのトラックでトラックリストを作成します。(Track tools > Create Track tool)



■ Trioの結果のトラックの名前のあたりでダブルクリックを行うと、テーブルが現れます。このテーブルと、トラックリストが連携しているため、興味の対象となる変異をテーブルで選ぶと自動的にトラックリストの該当箇所を見つけることができます。