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单体型分析PGD报告

**基本信息：**

|  |  |  |  |
| --- | --- | --- | --- |
| **样本序号** | **性别** | **亲缘关系** | **致病基因信息** |
| ALD150001 | 女 | 女方 | EX7:c.1762\_1763insCAC, p.Ile588delinsThrLeu Het |
| ALD150002 | 男 | 男方 | 未见异常 |
| 20150291-E2(2) | - | 胚胎 |  |
| 20150291-E3(2) | - | 胚胎 |  |

**检测方法：**

以ABCD1基因（NM\_000033.3 chrX: 152990323-153010216正向转录）编码区为目标区域，并在基因上下游选择若干SNP位点作为遗传标记，为鉴定样本性别，在Y染色体上选择10个特异位点，高通量测序检测SNP基因型，选择其中相应位点构建单体型。SNP位点分布见下表：

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **区域** | **基因下游** | | **基因上游** | | **Y染色体** |
| **1M外** | **1M内** | **1M内** | **1M外** |
| SNP数/个 | 15 | 71 | 76 | 85 | 10 |

**检测结果：**

|  |  |  |  |
| --- | --- | --- | --- |
| **样本序号** | **单体型检测结果** | **一代验证**  **c.1762\_1763insCAC** | **备注** |
| ALD150001 | 女性携带 |  |  |
| ALD150002 | 男性正常 |  |  |
| 20150291-E2(2) | 男性正常 | Normal |  |
| 20150291-E3(2) | 女性正常 | Normal |  |

**目标基因SNP位点统计：**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **样本序号** | **基因上游** | | | | **基因内部** | | **基因下游** | | | |
| **1M外** | | **1M内** | | **1M内** | | **1M外** | |
| **男方** | **女方** | **男方** | **女方** | **男方** | **女方** | **男方** | **女方** | **男方** | **女方** |
| ALD150001 | - | 2 | - | 30 | - | 0 | - | 33 | - | 2 |
| ALD150002 | 0 | - | 0 | - | 0 | - | 0 | - | 0 | - |
| 20150291-E2(2) | 0 | 2 | 0 | 29 | 0 | 0 | 0 | 32 | 0 | 2 |
| 20150291-E3(2) | 0 | 2 | 0 | 29 | 0 | 0 | 0 | 33 | 0 | 2 |

**高通量测序突变检测：**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **样本序号** | **chrX:153006148 c.1762\_1763insCAC** | | | |
| **Genotype** | **Depth** | **Ref** | **Alt** |
| ALD150001 | Het | 7646 | G(6678) | GCAC(968) |
| ALD150002 | Normal | 7689 | G(7689) | . |
| 20150291-E2(2) | Normal | 3173 | G(3173) | . |
| 20150291-E3(2) | Normal | 3657 | G(3657) | . |

**性别鉴定：**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Name** | **Chr** | **Start** | **End** | **ALD150001** | **ALD150002** | **20150291-E2(2)** | **20150291-E3(2)** |
| Y\_1 | chrY | 2655171 | 2655398 | 7 | 1865 | 3783 | 2 |
| Y\_2 | chrY | 2656620 | 2656793 | 0 | 147 | 0 | 0 |
| Y\_3 | chrY | 6634948 | 6635122 | 1 | 704 | 232 | 0 |
| Y\_4 | chrY | 7071902 | 7072117 | 0 | 231 | 250 | 0 |
| Y\_5 | chrY | 8866328 | 8866417 | 5 | 2914 | 504 | 0 |
| Y\_6 | chrY | 14829388 | 14829583 | 1 | 619 | 205 | 0 |
| Y\_7 | chrY | 15995816 | 15996043 | 2 | 305 | 0 | 0 |
| Y\_8 | chrY | 17920184 | 17920411 | 0 | 387 | 125 | 0 |
| Y\_9 | chrY | 19418727 | 19418942 | 0 | 265 | 29 | 0 |
| Y\_10 | chrY | 23282850 | 23283070 | 1 | 515 | 1225 | 2 |

**SNP单体型：**



|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Num** | **Name** | **Position** | **ALD150001** | |  | **ALD150002** |  | **20150291-E2(2)** |  | **20150291-E3(2)** | |
| **M0** | **M1** |  | **F0** |  | **M0** |  | **M0** | **F0** |
| 1 | 0\_m | 150995745 | T | C |  | T |  | T |  | T | T |
| 2 | 1\_m | 151333840 | T | C |  | C |  | T |  | T | C |
| 3 | 2 | 151758287 | C | C |  | G |  | C |  | C | G |
| 4 | 3 | 151809358 | A | A |  | G |  | A |  | A | G |
| 5 | 4\_m | 152036426 | T | C |  | C |  | T |  | T | C |
| 6 | 5\_m | 152048528 | C | T |  | C |  | C |  | C | C |
| 7 | 6\_m | 152073755 | T | C |  | C |  | T |  | T | C |
| 8 | 7\_m | 152091035 | A | T |  | T |  | A |  | A | T |
| 9 | 8\_m | 152091153 | A | C |  | C |  | A |  | A | C |
| 10 | 9\_m | 152170051 | G | A |  | A |  | G |  | G | A |
| 11 | 10\_m | 152190287 | C | T |  | T |  | C |  | C | T |
| 12 | 11 | 152215504 | G | G |  | A |  | G |  | G | A |
| 13 | 12 | 152228332 | A | A |  | G |  | A |  | A | G |
| 14 | 13\_m | 152357842 | C | T |  | C |  | C |  | ? | ? |
| 15 | 14 | 152369818 | T | T |  | C |  | T |  | T | C |
| 16 | 15\_m | 152416481 | G | C |  | G |  | G |  | G | G |
| 17 | 16\_m | 152416494 | A | G |  | A |  | A |  | A | A |
| 18 | 17 | 152463813 | G | G |  | A |  | G |  | ? | ? |
| 19 | 18\_m | 152469310 | C | T |  | C |  | C |  | C | C |
| 20 | 19\_m | 152469396 | A | T |  | T |  | A |  | A | T |
| 21 | 20 | 152500502 | G | G |  | A |  | G |  | G | A |
| 22 | 21\_m | 152508909 | C | T |  | C |  | C |  | C | C |
| 23 | 22\_m | 152569678 | G | A |  | G |  | G |  | G | G |
| 24 | 23\_m | 152609158 | G | C |  | G |  | G |  | G | G |
| 25 | 24\_m | 152632681 | T | C |  | T |  | T |  | T | T |
| 26 | 25\_m | 152670352 | A | G |  | A |  | A |  | A | A |
| 27 | 26\_m | 152677682 | C | A |  | A |  | C |  | C | A |
| 28 | 27\_m | 152769477 | C | T |  | T |  | C |  | C | T |
| 29 | 28\_m | 152769502 | G | A |  | A |  | G |  | G | A |
| 30 | 29\_m | 152787260 | G | A |  | A |  | G |  | G | A |
| 31 | 30\_m | 152794075 | T | G |  | G |  | T |  | T | G |
| 32 | 31\_m | 152799127 | C | G |  | G |  | C |  | C | G |
| 33 | 32\_m | 152814373 | T | C |  | C |  | T |  | T | C |
| 34 | 33\_m | 152815886 | T | C |  | C |  | T |  | T | C |
| 35 | 34\_m | 152833893 | A | G |  | G |  | A |  | A | G |
| 36 | 35\_m | 152839112 | A | G |  | G |  | ? |  | A | G |
| 37 | 36\_m | 152842741 | C | G |  | G |  | C |  | C | G |
| 38 | 37\_m | 152869143 | G | A |  | G |  | G |  | G | G |
| 39 | 38 | 152886255 | G | G |  | A |  | G |  | G | A |
| 40 | 39\_m | 152901900 | C | T |  | T |  | C |  | C | T |
| 41 | 40 | 152902768 | G | G |  | T |  | G |  | G | T |
| 42 | 41 | 153029226 | G | G |  | C |  | ? |  | ? | ? |
| 43 | 42 | 153031337 | C | C |  | A |  | C |  | C | A |
| 44 | 43 | 153031840 | C | C |  | G |  | ? |  | C | G |
| 45 | 44 | 153037172 | G | G |  | A |  | ? |  | ? | ? |
| 46 | 45\_m | 153037275 | C | T |  | T |  | ? |  | ? | ? |
| 47 | 46\_m | 153041544 | C | T |  | C |  | C |  | C | C |
| 48 | 47\_m | 153045202 | G | A |  | G |  | G |  | G | G |
| 49 | 48 | 153045438 | G | G |  | A |  | ? |  | ? | ? |
| 50 | 49 | 153045812 | C | C |  | T |  | ? |  | ? | ? |
| 51 | 50 | 153049391 | A | A |  | G |  | A |  | A | G |
| 52 | 51 | 153050047 | C | C |  | G |  | C |  | C | G |
| 53 | 52 | 153050984 | G | G |  | A |  | G |  | G | A |
| 54 | 53 | 153065865 | G | G |  | A |  | G |  | G | A |
| 55 | 54 | 153067110 | G | G |  | A |  | G |  | G | A |
| 56 | 55 | 153080522 | G | G |  | A |  | G |  | G | A |
| 57 | 56 | 153143775 | G | G |  | A |  | G |  | G | A |
| 58 | 57 | 153143918 | C | C |  | A |  | C |  | C | A |
| 59 | 58 | 153146400 | A | A |  | G |  | A |  | A | G |
| 60 | 59 | 153147271 | G | G |  | A |  | G |  | G | A |
| 61 | 60 | 153148238 | G | G |  | C |  | G |  | G | C |
| 62 | 61 | 153156390 | T | T |  | C |  | T |  | T | C |
| 63 | 62 | 153162037 | A | A |  | C |  | A |  | A | C |
| 64 | 63 | 153168624 | T | T |  | C |  | ? |  | ? | ? |
| 65 | 64 | 153176959 | T | T |  | A |  | T |  | T | A |
| 66 | 65 | 153189819 | C | C |  | T |  | C |  | C | T |
| 67 | 66 | 153196057 | T | T |  | C |  | ? |  | ? | ? |
| 68 | 67\_m | 153197202 | A | G |  | G |  | A |  | A | G |
| 69 | 68\_m | 153207545 | G | A |  | G |  | G |  | G | G |
| 70 | 69\_m | 153211538 | A | G |  | A |  | A |  | A | A |
| 71 | 70\_m | 153211652 | G | A |  | G |  | G |  | G | G |
| 72 | 71\_m | 153216036 | A | G |  | A |  | A |  | A | A |
| 73 | 72\_m | 153222835 | G | C |  | C |  | ? |  | ? | ? |
| 74 | 73\_m | 153225634 | C | G |  | C |  | C |  | C | C |
| 75 | 74\_m | 153227770 | A | G |  | A |  | A |  | A | A |
| 76 | 75\_m | 153239587 | G | T |  | T |  | ? |  | ? | ? |
| 77 | 76\_m | 153247722 | T | C |  | T |  | T |  | T | T |
| 78 | 77\_m | 153247745 | G | A |  | G |  | G |  | G | G |
| 79 | 78\_m | 153247954 | T | C |  | T |  | T |  | T | T |
| 80 | 79\_m | 153248248 | A | G |  | A |  | ? |  | A | A |
| 81 | 80\_m | 153296975 | A | G |  | A |  | A |  | A | A |
| 82 | 81\_m | 153387218 | T | G |  | T |  | T |  | T | T |
| 83 | 82\_m | 153387918 | A | G |  | A |  | A |  | A | A |
| 84 | 83\_m | 153390481 | G | A |  | G |  | G |  | G | G |
| 85 | 84\_m | 153546097 | C | T |  | C |  | C |  | C | C |
| 86 | 85\_m | 153558418 | A | G |  | A |  | A |  | A | A |
| 87 | 86\_m | 153595418 | T | C |  | T |  | T |  | T | T |
| 88 | 87\_m | 153602265 | T | C |  | T |  | T |  | T | T |
| 89 | 88\_m | 153609963 | C | T |  | C |  | C |  | C | C |
| 90 | 89\_m | 153612371 | G | C |  | G |  | G |  | G | G |
| 91 | 90\_m | 153660070 | T | C |  | T |  | T |  | T | T |
| 92 | 91\_m | 153880830 | C | A |  | A |  | C |  | C | A |
| 93 | 92\_m | 153887204 | T | C |  | C |  | T |  | T | C |
| 94 | 93\_m | 153888135 | A | G |  | G |  | A |  | A | G |
| 95 | 94\_m | 153903203 | T | A |  | A |  | T |  | T | A |
| 96 | 95\_m | 153903395 | A | G |  | G |  | A |  | A | G |
| 97 | 96\_m | 153904473 | T | C |  | C |  | T |  | T | C |
| 98 | 97\_m | 153908832 | A | G |  | G |  | A |  | A | G |
| 99 | 98\_m | 153948160 | C | T |  | T |  | C |  | C | T |
| 100 | 99\_m | 153952147 | C | T |  | T |  | C |  | C | T |
| 101 | 100\_m | 154285851 | G | C |  | C |  | G |  | G | C |
| 102 | 101\_m | 154355630 | C | T |  | T |  | C |  | C | T |

\*红色底色代表致病位点，“？”代表该位点未检测到，黄色底色代表该位点发生脱扣， 表示基因边界， 表示致病位点边界， 代表女方有效位点， 代表男方有效位点， 代表重组不确定区域。



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|  |  | 报告日期：2018年02月06日 |
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备注：此结果只作为检测结果，不能作为临床诊断报告。