

Supp. Figure S1. A genome-wide 10K SNP array of patients 1, 2 and 3 from the North American genetic isolate showed a homozygous region ~9.8 Mb on chromosome 17 shared by all three patients. The yellow indicates the number of contiguous homozygous SNPs in each chromosomal region, and the purple indicates the location score, which is a measure of the frequency of particular SNPs in the general population.

Loucks et al. Supplementary Figure 1

Loucks et al. Supplementary Figure 2

А				$\neg $			
D17S849	432 551		263 259	267 265			
D17S831	1 910 488		108 110	110 106			
D17S1828	3 810 467		108 <mark>106</mark>	106 102			
D17S938	6 249 300		255 <mark>256</mark>	<mark>256</mark> 256			
D17S1791	9 156 457		284 <mark>274</mark>	274 280			
D17S1852	10 515 507		307 295	295 303			
D17S799	13 170 971		192 192	192 192			
D17S1857	16 415 226		174 167	169 153			
				1	2	3	4
D17S849	432 551	263 267	259 265	259 267	259 267	259 261	/ /
D17S831	1 910 488	108 <mark>110</mark>	<mark>110</mark> 106	110 110	110 110	110 110	<mark>110</mark> 106
D17S1828	3 810 467	108 <mark>106</mark>	106 102	106 106	106 106	106 106	<mark>106</mark> 104
D17S938	6 249 300	255 <mark>256</mark>	256 256	256 256	256 256	256 256	<mark>256</mark> 256
D17S1791	9 156 457	284 <mark>274</mark>	<mark>274</mark> 280	274 274	274 274	274 274	274 292
D17S1852	10 515 507	307 295	195 303	295 295	295 295	307 295	307 299
D17S799	13 170 971	192 192	192 192	192 192	192 192	202 192	202 206
D17S1857	16 415 226	174 169	167 153	167 169	167 169	171 169	171 169

В

D17S849	432 551	259	261	1	1
D17S831	1 910 488	110	110	110	106
<i>DPH1</i> c.17T>A	1 933 465	Α	А	А	А
rs7215857	2 341 350	С	С	С	C
rs3213696	2 573 652	Т	Т	Т	T
rs3213697	2 573 858	Т	Т	Т	Т
rs769427	3 119 767	Т	Т	Т	Т
rs17174920	3 119 871	Т	Т	Т	T
rs12944745	3 448 272	Т	Т	Т	T
rs12945853	3 448 331	Т	Т	Т	Т
rs9911213	3 448 434	G	G	G	G
D17S1828	3 810 467	106	106	106	104
D17S938	6 249 300	256	256	256	256
D17S1791	9 156 457	274	274	274	292
D17S1852	10 515 507	307	295	307	299
D17S799	13 170 971	202	192	202	206
D17S1857	16 415 226	171	169	171	169

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Supp. Figure S2. Microsatellite mapping confirmed the homozygous region found on chromosome 17 in patients 1-3 and targeted SNP genotyping revealed that only a small ~1.9 Mb region containing the c.17T>A *DPH1* mutation is shared by all four patients from the North American isolate **A:** Microsatellite mapping confirming the homozygous region on chromosome 17 found from the 10K SNP array. The marker names are indicated on the left, followed by their

Mb positions according to NCBI build 37.1. For each individual, both microsatellite alleles are shown as the sequence length in base pairs. Markers in red represent regions that were homozygous as determined by the 10K SNP array on patients 1 and 2 their unaffected siblings. These homozygous regions were shared by patient 3 but not by patient 4. **B**: Targeted SNP genotyping revealed a small ~1.9 Mb homozygous region shared by all four patients between the *DPH1* c.17T>A variant and the D17S1828 microsatellite marker.