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## Data Article

# Data supporting the co-expression of *PDHA1* gene and of its paralogue *PDHA2* in somatic cells of a family



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## ABSTRACT

This article presents a dataset proving the simultaneous presence of a 5'UTR-truncated *PDHA1* mRNA and a full-length *PDHA2* mRNA in the somatic cells of a PDC-deficient female patient and all members of her immediate family (parents and brother).

We have designed a large set of primer pairs in order to perform detailed RT-PCR assays allowing the clear identification of both *PDHA1* and *PDHA2* mRNA species in somatic cells. In addition, two different experimental approaches were used to elucidate the copy number of *PDHA1* gene in the patient and her mother.

The interpretation and discussion of these data, along with further extensive experiments concerning the origin of this altered gene expression and its potential therapeutic consequences, can be found in "Complex genetic findings in a female patient with pyruvate dehydrogenase complex deficiency: null mutations in the *PDHX* gene associated with unusual expression

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of the testis-specific *PDHA2* gene in her somatic cells" (A. Pinheiro, M.J. Silva, C. Florindo, et al., 2016) [1].

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## Specifications Table

Subject area	<i>Biology</i>
More specific subject area	<i>Molecular Genetics</i>
Type of data	<i>Tables, figures</i>
How data was acquired	<i>Agarose gel electrophoresis after RT-PCR analyses quantitative real time PCR, microarray analyses, in silico analyses (BLAST software)</i>
Data format	<i>Raw, analyzed</i>
Experimental factors	<i>Genomic DNA and total RNA isolated from whole blood samples and fibroblast cultures</i>
Experimental features	<i>Genomic DNA was amplified by quantitative real time PCR and microarray analyses. Total RNA was reverse transcribed and amplified by semi-quantitative RT-PCR and by quantitative real time PCR using TaqMan assays. Alignment of sequences was performed using the BLAST software.</i>
Data source location	<i>Lisboa, Portugal</i>
Data accessibility	<i>Data provided within the manuscript and available in public databases (NCBI) in case of sequence alignment: GenBank accession numbers GenBank: NM_000284.3 (<i>PDHA1</i>) and GenBank: NM_005390.4 (<i>PDHA2</i>)</i>

## Value of the data

- These data, reporting on *PDHA2* gene expression in somatic cells, may trigger new research related to the activation of a paralogue gene as a therapeutic target to loss-of-function mutations.
- Data revealing the co-existence of both *PDHA1* and *PDHA2* mRNAs in somatic cells will be useful for future experiments addressing the impact between both isoforms in the assembly of a fully functional PDC.
- Data concerning gene copy number may assist the choice of the underlying methodology.
- These dataset may contribute for designing further experiments aiming the development of alternative therapies for metabolic disorders.

## 1. Data

The E1 rate-limiting enzyme of pyruvate dehydrogenase complex (PDC) is a heterotetramer ( $\alpha_2\beta_2$ ) and its  $\alpha$  subunit is encoded by *PDHA1* gene, located in X chromosome and presenting ubiquitous expression in somatic tissues. Nevertheless a paralogue gene exists, *PDHA2*, which is located in chromosome 4 and expressed only in spermatocytes and spermatids [2].

Table 1 shows the primers used for the amplification of the analyzed genes, according to the used methodology. Fig. 1 presents the results of *PDHA1* and *PDHA2* gene expression in somatic cells of the individuals under study and in controls. Fig. 2 displays the alignment of *PDHA1* and *PDHA2* mRNAs

**Table 1**

List of primers used in this study.

Primer	Sequence	Position
<b>cDNA amplification</b>		
<b>PDHA1 messenger</b>		
PDHA1-F	5'-AGCATCCCCTAATTTTGC-3'	+75 to +92
PDHA1-R	5'-CTTTAGTTCCTCCACTGG-3'	+989 to +1008
PDHA1-5'-F	5'-GGGCACCTGAAGGAGACTT-3'	-85 to -66
PDS1	5'-TGTGAGGAGTCGCCGCTGCC-3'	-37 to -18
PDSTr-F	5'-GCCACTGCCTGTGCTTCAT-3'	-17 to +2
PDSTr-R	5'-ACTCCATTCGGCGTACAGTCT-3'	+207 to +226
<b>PDHA2 messenger</b>		
PDHA2-F	5'-TGCCATCTACAGCACTCCGT-3'	-27 to -8
PDHA2-R	5'-CCTCCTTGAGTTGAGAACAC-3'	+1235 to +1254
<b>PDHX messenger</b>		
PXF2	5'-CTGCTGCGTTATCTTGTTGGCT-3'	+37 to +58
PXW2	5'-TGAGTGAATGTGCCACTGCATTG-3'	+812 to +835
PXP2	5'-CAATGCAGTGGGCACATCACTGA-3'	+812 to +835
PXR2	5'-TAACAACACTGAATCAACTAAGC-3'	+2060 to +2083
<b>Genomic DNA amplification</b>		
<b>PDHA1 gene</b>		
PDHA1-P1-F	5'-CCCTTGTTGCTTTGGTGT-3'	4383 to 4403
PDHA1-P1-R	5'-AGATTGCTCTGCTGACTACCG-3'	4762 to 4784
PDHA1-P2-F	5'-TGAGCATGCTGCTAATCTTCA-3'	4642 to 4682
PDHA1-P2-R	5'-CGGCGTGACAGAGCTGTAAT-3'	5114 to 5133
PDHA1-P3-F	5'-CTGGACGCCGTTCTGGTT-3'	4966 to 2983
PDHA1-P3-R	5'-GCGGAGGCGAAGTAAAGG-3'	4323 to 4340
PDHA1-P4-F	5'-TGCTTCATGAGGAAGATGCT-3'	5140 to 5159
PDHA1-P4-R	5'-AGGGTGCTTTGAACGAAG-3'	5526 to 5645
<b>PDHA2 gene</b>		
PDHA2-A-F	5'-GAGTAAGGAAAAGTGAATGTCA-3'	-841 to -819
PDHA2-A-R	5'-ATCCTGCTCCATAATGTGCC-3'	-200 to -181
PDHA2-B-F	5'-GCCATCAGGATAAATGTGGC-3'	-657 to -638
PDHA2-B-R	5'-CCCTTTCCCTGTAAACCC-3'	-322 to -303
PDHA2-C-F	5'-AACTCTCAGAAGTCTCATGTGCC-3'	-415 to -393
PDHA2-C-R	5'-ACGGAGTGCTGTAGATGGCA-3'	-27 to -8
PDHA2-D-F	5'-CAGGACCTGCCTCTATCACC-3'	-142 to +123
PDHA2-D-R	5'-AAACCGCAATGAATTTCTG-3'	+244 to +263
PDHA2-F-F	5'-GCATGGAATGAAGGCAGAT-3'	+212 to +231
PDHA2-F-R	5'-CCTCCTTGAGTTGAGAACAC-3'	+1298 to +1317
<b>PDHX gene</b>		
PX1F	5'-AGAGACCTAAAGGCCCGCT-3'	+5414 to +5433
PX1R	5'-AAGCAGGCCCTCAATCATAA-3'	+5751 to +5770
PX2F	5'-TGGGAATCTTTAGACTTTGGA-3'	+20,144 to +20,165
PX2R	5'-TGCTGAACCCAGAAAACCTT-3'	+20,531 to +20,550
PX3F	5'-CAACCCAGAAATAGCTACGGA-3'	+36,259 to +36,279
PX3R	5'-CACATTAATAAAGGAGGCAAAA-3'	+36,557 to +36,581
PX4F	5'-TGCAGTCATGGGGTTTACTT-3'	+46,205 to +46,225
PX4R	5'-ACAGCAACTTCTACGTGATG-3'	+46,549 to +46,570
PX5F	5'-GTGACCATCTGTGGGAGTCA-3'	+49,159 to +49,173
PX5R	5'-TTAITCAGAAAACAACCTTGCAT-3'	+49,549 to +49,573
PX6F	5'-TCACCTGCGTTTTCTGAAAAGT-3'	+55,435 to +55,455
PX6R	5'-GTGAGCCAAGATTGTGCCAT-3'	+55,779 to +55,798
PX7F	5'-TTCCACTGTGTGTTTAAACGGA-3'	+58,968 to +58,988
PX7R	5'-TTTCCTTAGCACAAATATACCCA-3'	+59,294 to +59,318
PX8F	5'-ACAAGTTTGAAGTTGTAATGGTCA-3'	+66,918 to +66,941
PX8R	5'-GAGGGAGATCAAACGATAGGA-3'	+67,178 to +67,198
PX9F	5'-TTTTCTGTAACCCGCTTGG-3'	+73,376 to +73,395
PX9R	5'-TCTCCCTTACACACACAA-3'	+73,700 to +73,719

**Table 1** (continued)

Primer	Sequence	Position
PX10F	5'–GGTACAAAATCAAATCAAGGCA–3'	+81,064 to +81,085
PX10R	5'–TTCAGATAAATGAAAGGCTGACA–3'	+81,315 to +81,337
PX11F	5'–ACGGAAGGGGACTTTGATT–3'	+83,725 to +83,744
PX11R	5'–TTGAGGACTAGGCAAGTCGG–3'	+84,031 to +84,050
<b>PDHA2 gene methylation analysis</b>		
CpGI-M-F	5'–ATAAATTAGTTAGTTTGGTTGCGT–3'	–188 to –164
CpGI-M-R	5'–ATAACGTCATTTAAAAAATTACGAA–3'	+74 to +98
CpGI-U-F	5'–ATAAATTAGTTAGTTTGGTTGTGT–3'	–188 to –64
CpGI-U-R	5'–ATAACATCATTTAAAAAATTACAAA–3'	+74 to +98
CpGII-F	5'–TGGAATTGAAGGTAGATTAGTTGTATAAAT–3'	+205 to +234
CpGII-R	5'–ATACCATTACCCCATAAAAATTCT–3'	+406 to +431
<b>Gene dosage analysis</b>		
<b>PDHA1 gene</b>		
PDHA1-exon7F	5'–AGGAGGCCCTTCTGTGCTTT–3'	11,341 to 11,359
PDHA1-exon7R	5'–CGGCCCCACCACAGGGTTCCT–3'	11,616 to 11,636
<b>PAH gene</b>		
PAH-exon1F	5'–GCTTTACTGTGCGGAGATCACAC–3'	5315 to 5339
PAH-exon1R	5'–CTTATGAAACCAGGAAGCAC–3'	5606 to 5625

showing that the specific primers were designed to anneal to regions with null or very low homology between the two genes, thus proving the simultaneous presence of both transcripts. Fig. 3 depicts the scheme of *PDHA1* mRNA with the localization of all the primers used to prove the presence of the 5'UTR truncated *PDHA1* mRNA detected in the family samples, and to localize the truncation point. Table 2 and Fig. 4 show the results of the two different methodologies used to evaluate *PDHA1* gene copy number: quantitative real time PCR (Table 2) and microarray analyses (Fig. 4).

## 2. Experimental design, materials and methods

### 2.1. Sample preparation

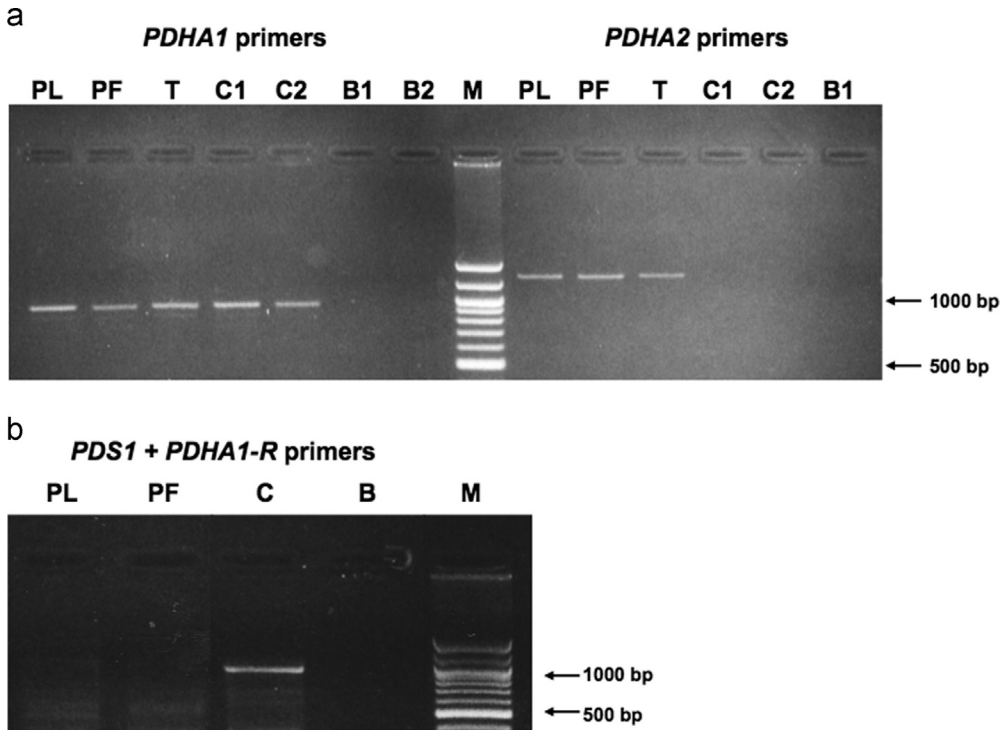
Lymphocytes were isolated from three independent peripheral blood samples obtained from the index case and her parents and brother, as well as from control individuals.

Patient's fibroblast cultures were established from a diagnostic skin biopsy and grown under standard conditions.

Positive controls for *PDHA2* gene expression were obtained from two different sources; a commercially available human testis total RNA sample (Clontech Laboratories Inc., Mountain View, CA, USA) and human testis specimens from eight cases requiring open testicular biopsy for the retrieval of testicular sperm for intracytoplasmic sperm injection [3].

### 2.2. Nucleic acids preparation

Genomic DNA, total RNA and cDNA were prepared according to standard methods and described in [1].



**Fig. 1.** RT-PCR analyses of PDH E1 $\alpha$  transcripts. (a) Using *PDHA1* and *PDHA2* specific primers. PL - patient lymphocytes; PF - patient fibroblasts; T - whole testis tissue; C1 and C2 - control lymphocytes; B1 without PCR control using whole testis total RNA; B2 - PCR control using no biological sample. M - 100 Base Pair Ladder (New England Biolabs). (b) Using forward *PDS1* primer and reverse *PDHA1* specific primer. PL - patient lymphocytes; PF - patient fibroblasts; C - control lymphocytes; B2 - PCR control using no biological sample. M - 100 Base Pair Ladder (New England Biolabs).

### 2.3. PCR of genomic DNA and cDNA

Amplification of the 11 individual exons of the *PDHA1* gene and related intron–exon boundaries were amplified using primers already published [4]. *PDHA1* and *PDHA2* cDNAs were amplified under conditions previously described [5] and using primers listed in Table 1, which were designed to annealing to regions displaying no homology between transcripts [6].

### 2.4. Evaluation of *PDHA1* and *PDHA2* expression and *PDHA1* gene dosage

*PDHA1* and *PDHA2* transcriptional levels were evaluated by quantitative real time RT-PCR under conditions previously described [1].

The copy number of *PDHA1* gene was evaluated by two methods, quantitative real time PCR and microarray analysis, as previously described [1].

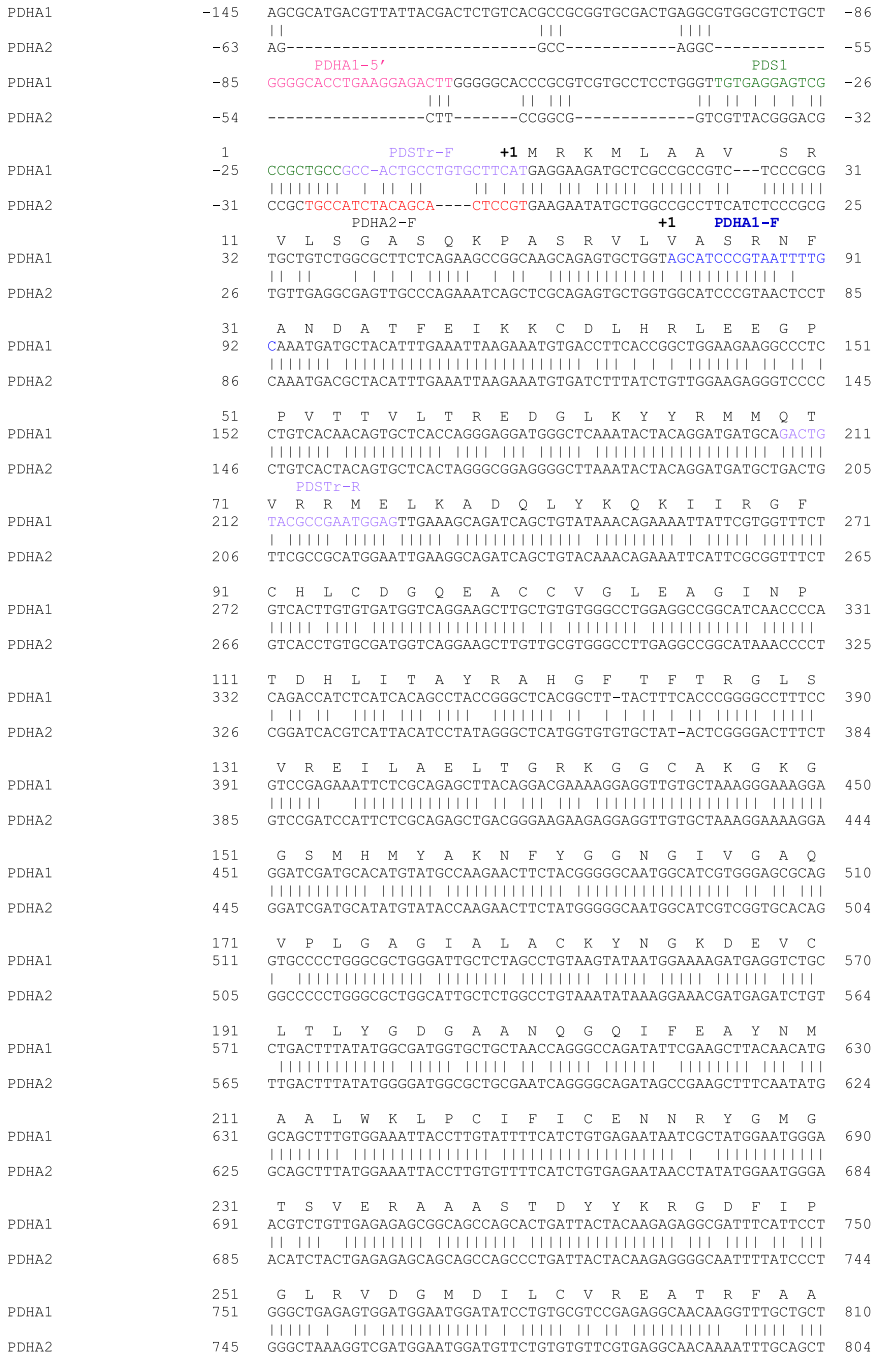


Fig. 2. Alignment of PDHA1 and PDHA2 cDNA sequences and primers' localization.

	271	A Y C R S G K G P I L M E L Q T Y R Y H	
PDHA1	811	GCCTATGTAGATCTGGGAAGGGGCCATCCTGATGGAGCTGCAGACTACCGTTACCAC	870
PDHA2	805	AACTACTGTAGATCTGGAAGGGGCCATACTGATGGAGCTGCAAACTACCGTTATCAT	864
	291	G H S M S D P G V S Y R T R E E I Q E V	
PDHA1	871	GGACACAGTATGAGTGACCCCTGGAGTCAGTTACCGTACACGAGAAGAAATTCAGGAAGTA	930
PDHA2	865	GGACACAGTATGAGTGATCCTGGAGTCAGTTATCGTACACGAGAAGAAATTCAGGAAGTA	924
	311	R S K S D P I M L L K D R M V N S N L A	
PDHA1	931	AGAAGTAAGAGTGACCCCTATTATGCTTCTCAAGGACAGGATGGTGAACAGCAATCTTGCC	990
PDHA2	925	AGAAGTAAGAGGGATCCTATAATAATCTCCAAGATAGAAATGGTAAACAGCAAGCTCGCC	984
		PDHA1-R	
PDHA1	331	S V E E L K E I D V E V R K E I E D A A	
	991	AGTGTGGGAAGAACTAAAGGAATTTGATGTGGAAGTGAAGGAGGATTGAGGATCGCTGCC	1050
PDHA2	985	ACTGTGGAAGAAATTAAGGAAATTTGGGGCTGAGGTGAGGAAAGAAATTTGATGATGCTGCC	1044
	351	Q F A T A D P E P P L E E L G Y H I Y S	
PDHA1	1051	CAGTTTCCCACGGCCGATCCTGAGCCACCTTTGGAAGAGCTGGGTACCACATCTACTCC	1110
PDHA2	1045	CAGTTTGTACTACTGATCCTGAGCCACATTTGGAAGAATTAGCCATCACATCTACAGC	1104
	371	S D P P F E V R G A N Q W I K F K S V S	
PDHA1	1111	AGCGACCCACCTTTTGAAGTTCGTGGTCCCAATCAGTGGATCAAGTTTAAGTCAGTCAGT	1170
PDHA2	1105	AGTGATTCATCTTTTGAAGTTCGTGGTGCAAATCCATGGATCAAGTTTAAGTCCGTCAGT	1164
PDHA1	1171	TAAGGGGAGGAGAAGGAGAGGTTATACCTTCAGGGGGCTACCAGACAGCTGTCTCAACT	1230
PDHA2	1165	TAAAGGGAGG-----CTAC-----	1178
PDHA1	1231	GGTTAAGGAGGAAGAAAACCCAGTCAATGAAATTCATGAAATTTCTGGAACTTCCATT	1290
		-----	
PDHA2		-----	
PDHA1	1291	AAGTGTGTAGATTGAGCAGGTAGTAATTCATGCAGTTTGTACATTAGTGCATTAAGA	1350
PDHA2	1179	--GTGTG-----AATT-----	1187
PDHA1	1351	TGAATTATGAGTGCTTAAAGATTATTTTGACTTAAAATAGTATACTTTGAACAAATAC	1410
PDHA2	1188	----TAT-----	1190
PDHA1	1411	TCTAATTATGAAAAGGAAGAACAATTCCTTGTATGCCTGTTTCCCTTGCCCCAGCCACC	1470
		-----	
PDHA2		-----	
PDHA1	1471	TTTTTGGGAGGAGACCATTATGGCGGGCCCTCACAGCATTTCACCAACCATAGCACCC	1530
PDHA2	1191	-----CAT-----CAG-----	1196
PDHA1	1531	ACCCCGAGCAGCGCTGGTGTGCAGCCTGTTCGCGTGACCATTTCCTCACAGATACAA	1590
PDHA2	1197	-----TCTCT-----CAA	1244
PDHA1	1591	TATTTATTATCAGGCAAGAGGACAGTTCCATTTTAAAATAAGACTTTTGTAAATCATTCCA	1650
PDHA2	1205	T-----GGA-----	1208
PDHA1	1651	ATTTTGTAAATCATTTCAAAGGCCACATAACTTAGTTTTCTCTACTTACACATTCAAGTATA	1710
		-----	
PDHA2		-----	
PDHA1	1711	AAATATGAAGCTATTTTCTGTTCATATCAAACATTAACATCAAGGCACATTTCGTATCAGTT	1770
PDHA2	1209	--ATG-----TTCAT-----GG-----	1218
PDHA1	1771	TTGTGTTTCTCAAATTTGAAGTACCATACCAGTTCTGAGGCAGTGTCCAGCTTCCATGTT	1830
PDHA2	1219	-----TCAAATTAAG-----	1229

Fig. 2. (continued)

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PDHA1      1831  TGTAAATACCCCTTGTTTGTTCACCATTCCAGCAAGTGCTGAAGGGTGTACTTTTTT 1890
                |||                               |||
PDHA2      1230  ---AAA-----CTG----- 1235
PDHA1      1891  GAGACAGGGTCGGGCTCTGTTGCCCAGGCTGGAGTGCAGTGGTGTGATCATGGCTCACTG 1950
PDHA2      1236  -----TGT-----CTCA----- 1243
                |||                               |||
                PDHA2-R
PDHA1      1951  CAGCCTCCACACCTCCTGGGCTCAAGCAATCCTCCCACCTCAGCCTCCTGCATAGCTGGG 2010
PDHA2      1244  -----AC-----TCAAG----- 1250
                ||           |||
PDHA1      2011  ACTACAAGTGAATTCCTAATATTCGGGAGTCAAACCAAGGCTCACTGTTTTCACAA 2070
PDHA2      1251  -----GAG----- 1254
                |||
PDHA1      2071  TACACACAGTTCTATGTTATAAAATACAGGTTTCAAAGAACTCAGGACAGTATTTAA 2130
                |||           |||           |||
PDHA2      1255  -----AATAA-----AACTCA-----TAA 1268
PDHA1      2131  AACCAAGTCTTAAACTATTAATTGAACAATGGCATTTTTAAATATGTAACACAGCGGAA 2190
                |||
PDHA2      1269  AACAA----- 1273
PDHA1      2191  TTCGTGTATACACTAACAGAAGCTTTAACAAAACATGTAGCGTGGTGGGACACTTGCCA 2250
PDHA2
PDHA1      2251  CAGCTTAGCTGATGGTATCAAGCCTTGCTTTGGTTTCTGAGGCCTCCTGAGCCCTTCT 2310
                |||           |||
PDHA2      1274  -----AAGCCTGT----- 1282
PDHA1      2311  GTACTGGGAGACCCGACTCCAGAGTCTGCAGAGGAGACCCTGGGAAACAAACACAG 2370
PDHA2
PDHA1      2371  CTGTCTTCAGAGTCAGTGCTTCAAGCCAACAGAGCTTAAACTGCAGTCCCTAATTTAAA 2430
                |||           |||
PDHA2      1283  -----AAGC-----ATTTA-- 1291
PDHA1      2431  AACCTAATGAAAATAAAAAACATTCTCCTCACATATGGAGGTGACGCTCGTGTCCCAGCAG 2490
PDHA2
PDHA1      2491  TAGTAGGACATGGCCTTAGAGGTACGTACCTGCAGAGAGCTGGCTATTTCAAATGACTCG 2550
                |||
PDHA2      1292  -----TTA----- 1294
PDHA1      2551  GGAACAAGAAGGCAGGCTGCAGTTTAAAGAAGGGGGTGGGTCCAGCGTGCAGGCACGCTT 2610
                |||
PDHA2      1295  -----AAAGA----- 1299
PDHA1      2611  GCCATGTGCCTCCACCCACTCCAGCCAGGCATTAATGGCAGGAGATTGGCCAGCTCTTC 2670
                |||
PDHA2      1300  -----GATT----- 1303
PDHA1      2671  TCTGTCACATTCCTATTTCTGACTTCTGCCTGGCTTTCAGTTTCTGCCACCTTGGCTT 2730
PDHA2
PDHA1      2731  TTTCCCAGCTTGAACCTAATAGAACTCCAGAGTTTGGGGGAGGCCAGCCCTTTGTTTT 2790
PDHA2
PDHA1      2791  CTGCTCTTGAAGCATATTCACACATAAAAAGTTGTATTCTCTTATACAACTGTTTGGAG 2850
PDHA2
PDHA1      2851  GCTCTTACCGTAGTCGAAGGTATCTTAGATCTCCTTAGTGATCTCATTAAGAATATCCG 2910
                |||
PDHA2      1304  -----ATTAA----- 1308
PDHA1      2911  AAAGTGATAACCCCTCTTCAACAATCTGAACAAAGATCAGATCCTTAAGAGCTGAGCAG 2970
                |||
PDHA2      1309  -----AAGAG----- 1313

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Fig. 2. (continued)

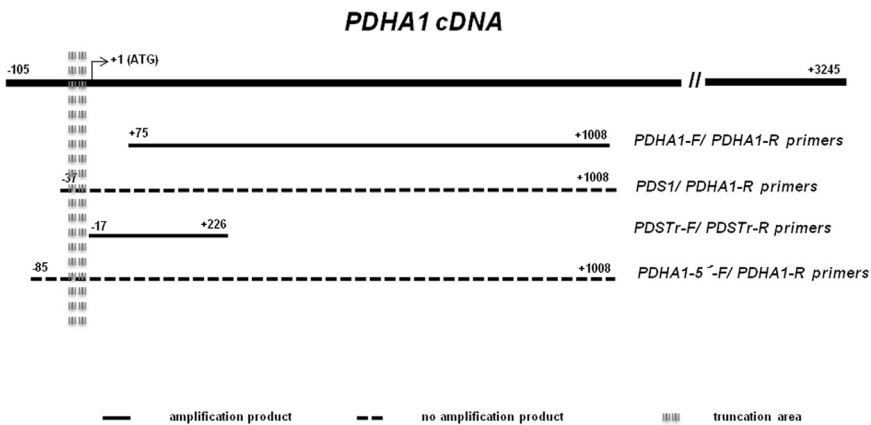


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PDHA1      2971  CTGTGTAACAACAGCATAAGAATTTCTTTGTTGTAATTTACCTTTTCAATTGTCTTTGC  3030
PDHA2      1314  -----ATTTG-----  1317
PDHA1      3031  ATCAGCTCCTTGCAGCCGCAACCAGTCTATAAGCTCTTTATCTGTCTCTGCCCGTAGGG  3090
PDHA2      -----
PDHA1      3091  GCCTGCTGGGTTCTCTGTAATACCTGTAACGATTGGCAATTTGTTATATATTAGTCTAAC  3150
PDHA2      -----
PDHA1      3151  CATAAAACTCTTCAAAGTAACCAGTTGGATTAATAAATGATTCAGAATGTAATGTGA  3210
PDHA2      -----
PDHA1      3211  TGTGAAAAAGAGATGAAAAAAAAAAAAAAAAAAAA  3245
PDHA2      1318  -----AAAGACA  1324

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Fig. 2. (continued)

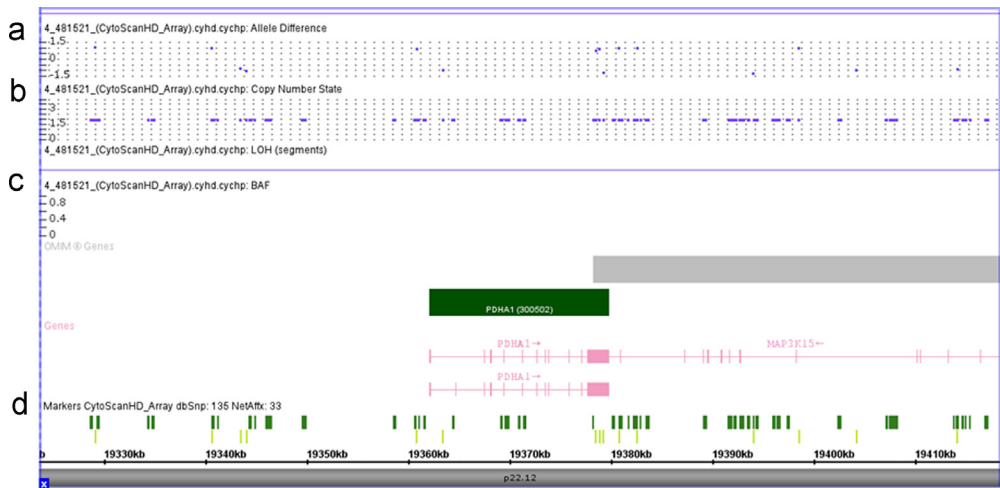


**Fig. 3.** Schematic representation of the *PDHA1* mRNA sequence showing the amplified versus non-amplified products in the RT-PCR analysis with the corresponding localization of the forward primers (PDHA1-5', PDS1, PDSTrF, PDHA1F) and reverse primers (PDHA1R and PDSTrR), as well as the identification of the predicted truncation point.

**Table 2**

Calculations for determining by qPCR the copy number of *PDHA1* gene using as reference the autosomal *PAH* gene.

<b>PDHA1 gene</b>				
Sample	Ave $\Delta$ Ct	$\Delta\Delta$ Ct	RQ ( $2^{-\Delta\Delta$ Ct})	Copy # ( $2 \times$ RQ)
Patient	0.26	0.91	0.5	1
Control Female 1	-0.65	0	1	2
Control Female 2	-0.33	0.32	0.8	2
Control Female 3	-0.59	0.06	0.9	2
Control Male 1	0.93	1.58	0.3	1
Control Male 2	-0.23	0.42	0.7	1
Control Male 3	-0.01	0.64	0.6	1



**Fig. 4.** Detailed view of the *PDHA1* region on chromosome X. (a) Allele difference and (b) copy number state showing absence of big deletions involving the gene. (c) OMIM genes: *PDHA1* (dark green horizontal bar) and *MAP3K15* (gray horizontal bar). Intron - horizontal pink lines; Exon - vertical pink bars. (d) Markers present in *PDHA1* region. Dark green - non-polymorphic probes; Light green - SNP, single nucleotide polymorphism. (For interpretation of the references to color in this figure legend, the reader is referred to the web version of this article).

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## Transparency document. Supporting information

Transparency data associated with this article can be found in the online version at <http://dx.doi.org/10.1016/j.dib.2016.08.029>.

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