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Rat uroporphyrinogen decarboxylase cDNA: nucleotide sequence and comparison to human uroporphyrinogen decarboxylase

Marc Romana, Philippe Le Boulch and Paul-Henri Roméo

Unité INSERM U.91, Hôpital Henri Mondor, 94010 Créteil, France
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Uroporphyrinogen decarboxylase (URO-D, E.C. 4.1.1.31) is a cytosolic enzyme involved in the heme biosynthetic pathway. In humans, a dominantly inherited disorder, Porphyria Cutanea Tarda (P.C.T.) is caused by a partial deficiency of URO-D activity (1) and a recent report (2) has shown a correlation between an amino acid substitution and the decrease in URO-D activity in a patient homozygous for the disease. However, this substitution was not found in unrelated heterozygous patients. We previously cloned the rat cDNA coding for uroporphyrinogen decarboxylase (3) and we present here its complete sequence. The nucleotide and amino acid sequence homologies between human (4) and rat uroporphyrinogen decarboxylase are 85% and 90% respectively. The amino acid substitutions are shown in Table I and indicate the protein regions which are not good candidates for the search for mutations in patients with PCT.

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CGAACGGCTTGGGACTCCAGAATTTCCCGGAGCTGAAGAATGACACGTTCTTGAGAGCAG   60
CCTGGGGAGAGGAAACAGACTATACTCCTGTTTGGTGCATGAGACAAGCAGGCCGCTACT  120
TACCAGAGTTTAGGGAAACCAGGGCTGCCCAGGACTTCTTCAGCACCTGTCGATCTCCTG  180
AGGCTTGCTGTGAACTGACTCTGGAGCCAGTGCGGAGGTTTCCTTTGGATGCTGCTATAA  240
TTTTCTCTGACATCCTTGTTGTACCCCAGGCATTGGCTATGGAGGTGACCATGGTACCTG  300
GCAAAGGACCCAGCTTTCCAGAGCCATTAAGAGAAGAGCGGGACTTAGAGCGTCTACGGG  360
ATCCAGCAGCAGTGGCTTCAGAGTTAGGCTATGTGTTCCAAGCCATCACCCTTACCCGAC  420
AACAGCTGGCTGGACGTGTGCCACTGATTGGCTTTGCTGGTGCTCCGTGGACCCTGATGA  480
CGTACATGGTTGAAGGCGGCAGTTTCAAGACCATGGCTCAGGCCAAGCGATGGCTCTATC  540
AGAAGCCAGTGGCCAGTCACAAGCTGCTTGGCATACTCACTCATGCTCTGGTCCCATATC  600
TAATAGGACAAGTAGCTGCTGGTGCTCAGGCATTACAGCTCTTTGAGTCCCACGCAGGAC  660
ATCTTGGCTCCGAGCTCTTCAGCAAGTTTGCACTGCCTTACATCCGTGATGTGGCCAAGC  720
GAGTGAAGGCTGGGTTGCAGAAGGCGGGCCTGACCAGGATGCCCATGATCATCTTTGCCA  780
AGGATGGACATTTTGCCCTGGAAGAGCTGGCCCAGGCTGGCTATGAGGTAGTTGGACTTG  840
ACTGGACAGTGGCTCCAAAGAAAGCCCGGGAACCTGTTGGAAAGACAGTGACTCTGCAGG  900
GGGAACTGGATCCCTGTGCCCTGTATGCATCTGAGGAAGAGATTGGTCGACTGGTGCAGC  960
AGATGCTGAATGACTTTGGGCCACAGCGCTACATTGCTAACCTAGGGCATGGGCTTTACC 1020
CTGACATGGACCCAGAACACGTAGGAGCCTTTTTGGATGCAGTACACAAACACTCACGCC 1080
TGCTTCGACAGAATTGAGTATGTGCTTTTCTGCTCAAGTACCACCGACACAGATTGTTTC 1140
CAGGAGAATAAAACTTCCAGAAACTTCCA                                 1200
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A. A. POSITION	7	9	23	72	94	102	115	124	125	143	169	170	171	183	185	189	192
HUMAN	P	G	W	L	G	S	Q	E	V	Q	G	S	S	R	Q	Q	R
RAT	L	N	T	V	A	G	R	A	A	R	S	F	K	K	V	K	G

196	203	207	225	226	229	242	246	248	249	253	264	265	293	303	318	321	325	330	353
D	V	V	P	Q	N	Q	R	R	E	A	P	V	K	N	Q	K	D	H	V
H	I	A	S	E	S	R	G	Q	K	T	R	M	P	E	R	Q	N	Q	L

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REFERENCES

1. Kushner, J., Barbuto, A.J. and Lee, G.R. (1976) J. Clin. Invest. 58:1089-1097.
2. De Verneuil, H., Grandchamp, B., Beaumont, C., Picat, C. and Nordmann, Y. (1986) Science 234:732-734.
3. Roméo, P.H., Dubart, A., Grandchamp, B., De Verneuil, H., Rosa, J., Nordmann, Y. and Goossens, M. (1984) Proc. Natl. Acad. Sci. USA 81:3346-3350.
4. Roméo, P.H., Raich, N., Dubart, A., Beaupain, D., Pryor, M., Kushner, J., Cohen-Solal, M. and Goossens, M. (1986) J. Biol. Chem. 261:9825-9831.