Clinical aspects and pathogenesis of congenital dyserythropoietic anemias: from morphology to molecular approach

Achille Iolascon,[1,2] Maria Rosaria Esposito,[2] and Roberta Russo[1,2]

[1]Department of Biochemistry and Medical Biotechnologies, University of Naples Federico II, Naples; and [2]CEINGE Biotecnologie Avanzate, Naples, Italy

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Online Supplementary Table 1S. Mutational spectrum of *CDAN1/CDAN2*

Gene	Exon	Nucleotide change°/*	Legacy name[a/b]	Reference
CDAN1	*missense*			
(54 cases)	2	c.156 C>G	F52L	(Ref. 2)
	6	c.1078 T>C	F360L	(Ref. 4)
	12	c.1796 A>G	N599S	(Ref. 1)
	14	c.2015 C>T	P672L	(Ref. 1)
	14	c.2062 C>T	R688W	(Ref. 2)
	14	c.2069 T>C	V690A	(Ref. 5)
	14	c.2092 G>A	E698K	(Ref. 1)
	14	c.2140 C>T	R714W	(Ref. 1)
	14	c.2173 C>T	R725W	(Ref. 4)
	15	c.2248 G>T	G750C	(Ref. 4)
	19	c.2602 T>A	F868I	(Ref. 1)
	19	c.2605 G>A	V869M	(Ref. 1)
	20	c.2650 A>G	T884A	(Ref. 2)
	24	c.3107 C>A	S1036F	(Ref. 1)
	24	c.3124 C>T	R1042W	(Ref. 1)
	24	c.3128 A>T	D1043V	(Ref. 1)
	24	c.3194 G>A	R1065Q	(Ref. 4)
	26	c.3389 C>T	P1130L	(Ref. 1)
	nonsense			
	14	c.2044 C>T	R682X	(Ref. 1)
	18	c.2539 C>T	Q847X	(Ref. 2)
	23	c.2992 C>T	R998X	(Ref. 1)
	27	c.3547 C>T	Q1183X	(Ref. 4)
	splicing			
	8	c.1367+1 G>A		(Ref. 2)
	12	c.1860+5 G>A		(Ref. 2)
	21	c.2868+2 insCCG		(Ref. 4)
	28	c.3558 del -10 to +31		(Ref. 4)
	frameshift			
	23	c.3024 insTT	G1008Gfs23X	(Ref. 4)
	24	c.3145 insT	S1049Ffs25X	(Ref. 4)
	small deletion			
	6	c.1117_19delGTT	V373del	(Ref. 3)
	12	c.1789_1791delGAG	E597del	(Ref. 4)
CDAN2				
(134 cases)	missense			
	2	c.40 C>T	R14W	(Ref. 7)
	2	c. 53 G>A	R18H	(Ref. 7)

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2	c.74 C>A	P25H	(Ref. 13)
2	c.197 G>A	C66Y	(Ref. 8)
4	c.325 G>A	E109K	(Ref. 7)
6	c.640 C>T	Q214X	(Ref. 12)
7	c.716 A>G	D239G	(Ref. 7)
8	c.938 G>A	R313H	(Ref. 7)
8	c.953T>C	I318T	(Ref. 7)
9	c.1043 A>C	D348A	(Ref. 6)
10	c.1157 A>G	Q386R	(Ref. 7)
11	c.1254 T>G	I418M	(Ref. 10)
11	c.1307 C>T	S436L	(Ref. 10)
12	c.1385 A>G	Y462C	(Ref. 7)
13	c.1453 A>G	T485A	(Ref. 12)
13	c.1489 C>T	R497C	(Ref. 7)
13	c.1508 G>A	R503Q	(Ref. 9)
14	c.1571 C>T	A524V	(Ref. 7)
14	c.1588 C>T	R530W	(Ref. 7)
14	c.1589 G>A	R530Q	(Ref. 9)
14	c.1654 C>T	L552F	(Ref. 10)
15	c.1685 A>G	Y562C	(Ref. 9)
15	c.1733 T>C	L578P	(Ref. 10)
15	c.1735 T>A	Y579N	(Ref. 10)
16	c.1808 C>T	S603L	(Ref. 6)
16	c.1832 G>C	R611P	(Ref. 10)
16	c.1858 A>G	M620V	(Ref. 10)
17	c.1910 T>G	V637G	(Ref. 12)
17	c.1968 T>G	F656L	(Ref. 9)
18	c.2101 C>T	R701C	(Ref. 6)
18	c.2129 C>T	T710M	(Ref. 11)
19	c.2166 A>C	K723Q	(Ref. 9)
19	c.2180 C>T	S727F	(Ref. 12)
20	c.2270 A>C	H757P	(Ref. 10)
nonsense			
2	c.71 G>A	W24X	(Ref. 13)
3	c.235 C>T	R79X	(Ref. 7)
5	c.367 C>T	R123X	(Ref. 10)
5	c.568 C>T	R190X	(Ref. 6)
6	c.649 C>T	R217X	(Ref. 7)
7	c.790 C>T	R264X	(Ref. 7)
8	c.970 C>T	R324X	(Ref. 7)
9	c.1015 C>T	R339X	(Ref. 10)
10	c.1201C>T	R401X	(Ref. 7)
14	c.1603 C>T	R535X	(Ref. 10)
14	c.1648 C>T	R550X	(Ref. 9)
14	c.1660 C<T	R554X	(Ref. 6)
splicing			
2	c.221+31 A>G		(Ref. 10)
3	c.279+3 A>G		(Ref. 10)
6	c.689+1 G>A		(Ref. 7)
9	c.1109+1 G>A		(Ref. 10)
9	c.1109+5 G>A		(Ref. 10)
19	c.2149-2 A>G		(Ref. 10)
frameshift			
3	c.222-817_366+4242del	Q75EfsX7	(Ref. 7)
5	c.387(del G)	L129LfsX26	(Ref. 10)

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5	c.428_428delAinsCG	D143AfsX35	(Ref. 6)
9	c.1063delG	D355IfsX8	(Ref. 7)
16	c.1821delT	H608IfsX7	(Ref. 6)
17	c.1962-64 (delT)	T654TfsX13	(Ref. 9)
19	c.2150 (delC)	A717VfsX7	(Ref. 10)
small deletion			
16	c.1857_1859delCAT	I619del	(Ref. 10)

*° CDAN1: the nucelotides are numbered from the A of the ATG initiation codon (ENST00000356231). All mutations have been revised according to the rules of nomenclature of the HGVS database (http://www.hgvs.org/mutnomen/). * CDAN2: the nucelotides are numbered from the A of the ATG initiation codon (ENST00000377475) [a]CODANIN-1: Accession number: Q8IWY9 (UniProtKB/Swiss-Prot). [b]SEC23B: Accession number: Q15437 (UniProtKB/Swiss-Prot).*

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