

Animal models of Diamond-Blackfan anemia: updates and challenges

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Suppl. Table-1: Genetic alternations and clinical manifestations in DBA patients

Gene (Protein)	% in DBA*	Alternation	Age at diagnosis	Hematopoiesis			Malformation**	Reference	
				Anemia	Elevated HbF	Elevated eADA		Author	Year
RPS19 (eS19)	25-30%	G120A	2 months	Yes	N/A***	N/A	No	Matsson et al	1999
		329delG	1 year						
		C302T	10 months						
		160insCT	15 months	Yes	Yes	Yes	Yes	Draptchinskaja et al	1999
		C302T	1 month						
		c.172+1G>A	8 months						
c.185G>A	Birth-23 yeas	Yes	Yes	Yes	Yes	Cole et al	2022		
RPL11 (uL5)	5-20%	c.59T>A, c.357T>G	Birth	Yes	Yes	N/A	Yes	Cmejla et al	2009
		Small indel	Birth-2 months	Yes	N/A	Yes	Yes	Quarello et al	2020
		Nonsense, splice site, small indel	1 month-21 yrs	Yes	N/A	N/A	Yes	Gazda et al	2008
		Nonsense, missense, splice site, small indel	Birth-10 months	Yes	N/A	N/A	Yes	Gazda et al	2008
RPL5 (uL18)	7-12%	Small indel	Birth-24 months	Yes	N/A	Yes	Yes	Quarello et al	2020
		Point mutations, splice site	Birth	Yes	N/A	N/A	Yes	Cmejla et al	2009
		Small indel	2 months	Yes	N/A	N/A	Yes	Gazda et al	2008
RPL36 (eL36)	<1%	Small indel	2 months	Yes	N/A	N/A	Yes	Gazda et al	2008
RPS26 (eS26)	6.6-9%	9 distinct mutations	6 weeks-8 months	Yes	N/A	N/A	No	Doherty et al	2010
RPS10 (eS10)	1-3%	3 distinct mutations	Birth-12 years	Yes	N/A	N/A	No	Doherty et al	2010
RPS7 (eS7)	~1%	Missense mutation	Adulthood	Yes	N/A	Yes	No	Volejnikova et al	2020
		Splice site variant	1st year	Yes	N/A	N/A	Yes	Akram et al	2020
		c.76-1G>T	Early infancy	Yes	N/A	Yes	Yes	Ichimura et al	2017
		Splice site in intron 1	1st year	Yes	N/A	N/A	Yes	Smetanina et al	2015
RPL35A (eL33)	2-3%	Gene deletion	4 months	Yes	N/A	Yes	Yes	Quarello et al	2020
		Allelic del, c.304 C>T, c.82_84delCTT	2-14 months	Yes	N/A	Yes	Yes	Farrar et al	2008
		Large deletion	Birth	Yes	Yes	Yes	Yes	Gianferante et al	2021
RPS24 (eS24)	2-3%	Nonsense mutation in Exon 4	N/A	Yes	No	Yes	N/A	Gazda et al	2006
RPS17 (eS17)	1-3%	Nonsense mutation	Birth, 11 months	Yes	N/A	Yes	Yes	Gerrard et al	2013
		c.2T>G	6 months	Yes	N/A	Yes	Yes	Cmejla et al	2007
		c.1A>G	5 months	Yes	N/A	N/A	Yes	Song et al	2010
RPL9 (uL6)	<1%	c.-2+1G>C, c.59T>C; c.59T>C	6 months	Yes	Yes	No	Yes	Lezzerini et al	2020
RPL15 (eL15)	~1%	Deletion of intron 3 and exon 4	Birth	Yes	N/A	N/A	Yes	Landowski et al	2013
		c.242dupA, c.85C>T, c.29T>C, c.458A>C	Prenatal to 6 months	Yes	Yes	Yes	Yes	Wlodarski et al	2018
RPS29 (uS14)	~1%	p.I31F, p.I50T	3 months-9 years	Yes	No	Yes	No	Mirabello et al	2014
RPS20 (uS10)	rare	c.251T>A, c.251T>G	1st month	Yes	N/A	Yes	No	Bhar et al	2020
RPL26 (uL24)	Rare	C.120_121delGA (nonsense mutation)	Birth	Yes	Yes	N/A	Yes	Gazda et al	2012
RPS27A (eS31)	Rare	c.169T>C	15 months	Yes	N/A	N/A	Yes	Gazda et al	2008
RPS28 (eS28)	Rare	c.1A>G	Infancy	Yes	N/A	N/A	Yes	Gripp et al	2014
GATA1	<1%	chX: 48,649,736 G>C; chX: 48,649,736-48,649,737Gdel	Birth, 1.6 months	Yes	Yes	Yes	N/A	Sankaran et al	2012
		c.220G>C	3 months	Yes	Yes	Yes	No	Klar et al	2014
		c.2T>C	27 and 28 yrs	Yes	N/A	N/A	N/A	Ludwig et al	2014
		c.2T>C	9 months	Yes	N/A	Yes	No	Parrella et al	2014
		c.2T>C, c.220+2T>C	7 months and 5 yrs	Yes	Yes	Yes	No	van Dooijeweert et al	2022
TSR2	Rare	c.191A>G	10 months	Yes	Yes	Yes	Yes	Gripp et al	2014
HEATR3	Rare	c.1751G>A, c.1337G>A, c.399+1G>T, c.719C>T, c.400T>C	5-20yrs	Yes	N/A	N/A	Yes	O'Donohue et al	2022
EPO	One case	chr7:100320704 G>A	1st year	Yes	N/A	N/A	N/A	Kim et al	2017

* Resource from <https://www.ncbi.nlm.nih.gov/books/NBK7047/> and <https://www.ncbi.nlm.nih.gov/pubmed/34889440>

**Malformation

*** N/A: Not available