Clinical features of *DDX41* mutation-related diseases: a systematic review with individual patient data

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Supplementary contents

Supplementary figure 1 Forest plot of *DDX41* incidence (see below)

For supplementary tables, please check extra excel files (.xlsx).

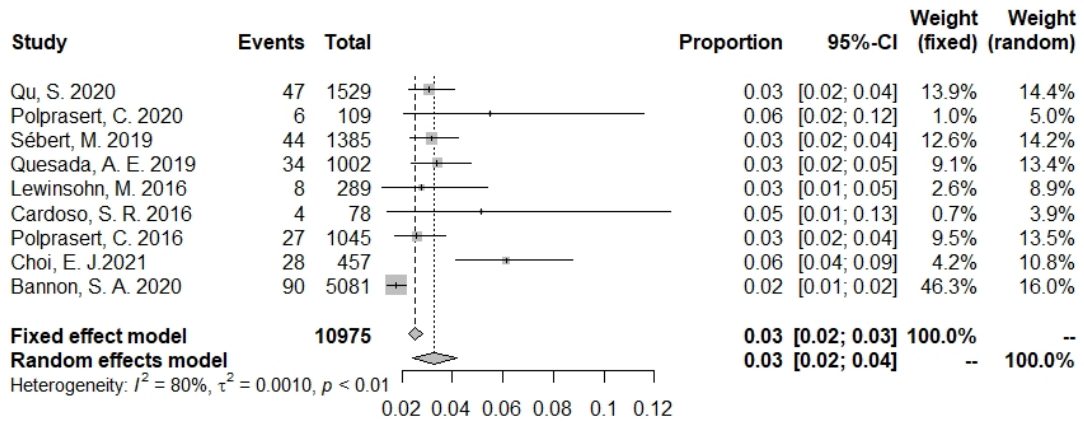Supplementary table 1 Reasons to include all the 30 articles

Supplementary table 2 Individual patient-level dataset

Supplementary table 3 Quality evaluation of 20 articles in the quantitative analysis

Supplementary table 4 Comparisons of AML, MDS and other diagnoses

Supplementary table 5 Comparisons of familial and sporadic cases

Supplementary table 6 Comparisons of carrying p.R525H and other somatic variants

Study	Events	Total	Proportion	95%-CI	Weight (fixed)	Weight (random)
Qu, S. 2020	47	1529	0.03	[0.02; 0.04]	13.9%	14.4%
Polprasert, C. 2020	6	109	0.06	[0.02; 0.12]	1.0%	5.0%
Sébert, M. 2019	44	1385	0.03	[0.02; 0.04]	12.6%	14.2%
Quesada, A. E. 2019	34	1002	0.03	[0.02; 0.05]	9.1%	13.4%
Lewinsohn, M. 2016	8	289	0.03	[0.01; 0.05]	2.6%	8.9%
Cardoso, S. R. 2016	4	78	0.05	[0.01; 0.13]	0.7%	3.9%
Polprasert, C. 2016	27	1045	0.03	[0.02; 0.04]	9.5%	13.5%
Choi, E. J.2021	28	457	0.06	[0.04; 0.09]	4.2%	10.8%
Bannon, S. A. 2020	90	5081	0.02	[0.01; 0.02]	46.3%	16.0%
Fixed effect model		**10975**	**0.03**	**[0.02; 0.03]**	**100.0%**	**--**
Random effects model			**0.03**	**[0.02; 0.04]**	**--**	**100.0%**

Heterogeneity: $I^2 = 80\%$, $\tau^2 = 0.0010$, $p < 0.01$

0.02 0.04 0.06 0.08 0.1 0.12

Supplementary figure 1 Forest plot of *DDX41* incidence

The pooled incidence of *DDX41* mutation among 10975 screened patients with myeloid neoplasms from 9 case series was 3.3% ([95%CI 2.4-4.2%], I2=79.7%, p<0.01).