

Pulmonary Manifestations of GATA2 Deficiency

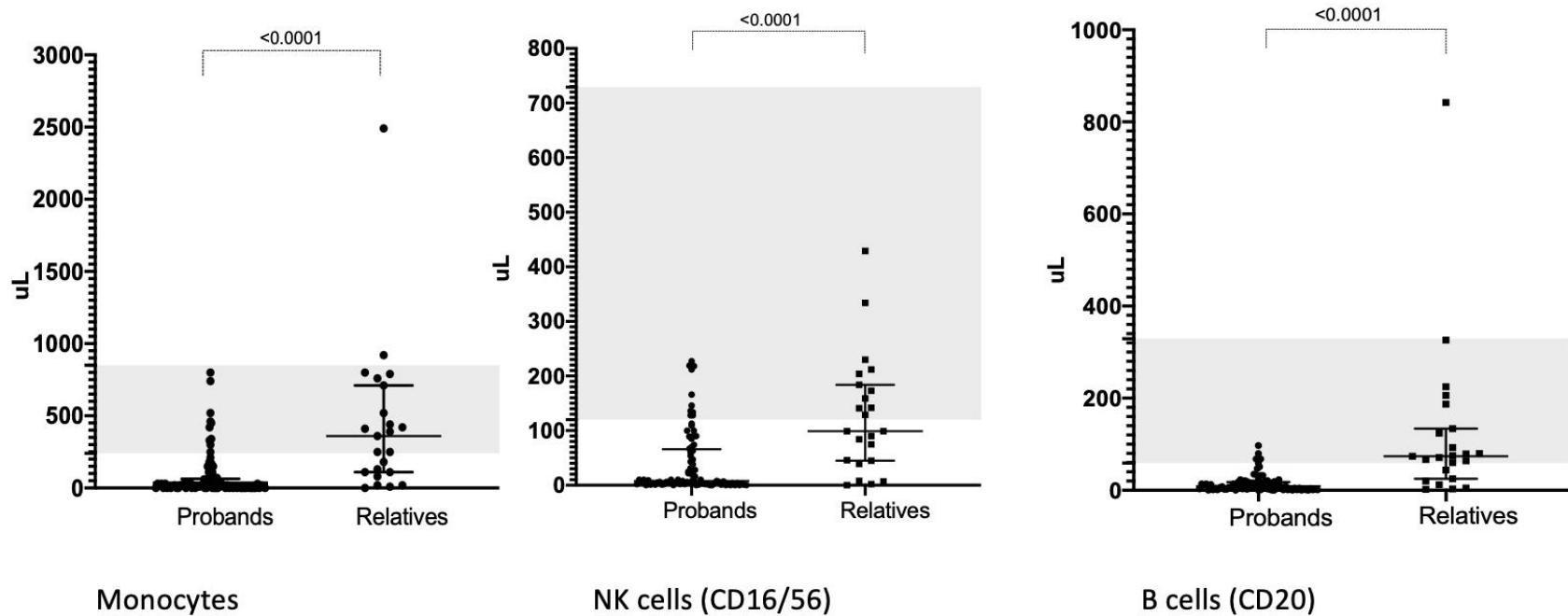
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e-Figure 1. Immunological profile in probands and relatives at the pulmonary evaluation. expressed in median and interquartile. Grays are normal values.



Immunological profile differences between probands and relatives (median IQ)
P value= $p<0.001$

e-Table 1. Infections in GATA2 patients.
Frequency of different organism in GATA2 deficient with and without pulmonary alveolar proteinosis.

Organisms isolated	Species	Patients without PAP (n=113)	Patients with PAP (n=11)
<i>Mycobacteria</i> *		33 (30%)	9 (81%)
	<i>M. kansasii</i>	13(11%)	2(2%)
	<i>M. avium</i> complex	13(11%)	5(5%)
	<i>M. abscessus</i>		1(1%)
	<i>M. bolletti</i>	1(<1%)	
	<i>M. fortuitum</i>	5(5%)	2(2%)
	<i>M. szulgai</i>	1(<1%)	
	No speciation	2(2%)	2(2%)
	<i>M. tuberculosis</i>	3(3%)	
Fungi		21 (18%)	6 (7%)
	<i>Aspergillus sp</i> [†]	2(2%)	4(4%)
	<i>Histoplasma</i>	3(3%)	2(2%)
	<i>Candida</i> [‡]	13(11%)	
	<i>Blastomyces</i>	2(2%)	
Viruses		77 (68%)	9 (100%)
	HSV	8 (7%)	
	HPV	59 (52%)	7 (8%)
	Varicella	7 (6%)	1 ^{**} (1%)
	CMV	4 (3%)	2 (2%)
	EBV	12 (10%)	

* More than one species isolated in some patients.

† *A. fumigatus* and one case of *A. neosartorya*

‡ Noninvasive (oral or vulvovaginitis)

|| Including warts

** Varicella-zoster infection

e-Table 2. Heat map of radiographic findings in GATA2 deficiency probands and relatives grouped according to mutation type.

ID	Published	Patients	Age	Nodules	Reticular	Paraseptal Emphysema	Ground-glass opacity	Consolidation	Crazy paving	Mutation type
378.I.1	*	Proband	12							Missense
141.I.1	*	Proband	16							Missense
15.I.1	*	Proband	16							Missense
34.II.2	*	Proband	16							Missense
351.I.1	*	Proband	17							Missense
1.IV.1		Proband	17							Missense
277.I.1	*	Proband	18							Missense
357.II.1	*	Proband	18							Missense
360.I.1	*	Proband	20							Missense
19.II.1	*	Proband	21							Missense
382.I.2	*	Proband	22							Missense
270.I.1	*	Proband	22							Missense
383.II.1	*	Proband	22							Missense
400		Proband	22							Missense
9.II.1	*	Proband	22							Missense
18.I.2	*	Proband	23							Missense
51.III.1	*	Proband	24							Missense
42.I.1	*	Proband	24							Missense
312.I.1	*	Proband	24							Missense
256.I.1	*	Proband	25							Missense
324.I.1	*	Proband	27							Missense
281.I.1	*	Proband	28							Missense
12.I.1	*	Proband	28							Missense
349.IV.5	*	Proband	30							Missense
339.I.1	*	Proband	31							Missense
146.I.1	*	Proband	32							Missense
351.I.1	*	Proband	32							Missense
31.II.1	*	Proband	33							Missense
21.II.1	*	Proband	33							Missense
37.I.1	*	Proband	35							Missense
17.I.1	*	Proband	36							Missense
402		Proband	37							Missense
35.III.3	*	Proband	37							Missense
365.I.1	*	Proband	38							Missense
10.I.1	*	Proband	38							Missense
216.I.1	*	Proband	38							Missense
2.II.3	*	Proband	40							Missense
384.I.1	*	Proband	40							Missense
5.I.1	*	Proband	41							Missense
384.I.1	*	Proband	41							Missense
218.I.1	*	Proband	44							Missense
1.II.1	*	Proband	45							Missense
52.I.4	*	Proband	45							Missense
24.I.1	*	Proband	47							Missense
1.II.5	*	Proband	48							Missense
333.II.3	*	Proband	52							Missense
31.I.1	*	Proband	55							Missense
301.I.1	*	Proband	55							Missense
40.I.1	*	Proband	55							Missense
349.II.4	*	Proband	66							Missense
405		Proband	9							Null
406		Proband	14							Null
47.II.1	*	Proband	18							Null
32.I.1	*	Proband	18							Null
50.I.2	*	Proband	18							Null
407		Proband	18							Null
408	*	Proband	19							Null
409	*	Proband	19							Null
33.III.1	*	Proband	20							Null
291.I.1	*	Proband	21							Null
410	*	Proband	21							Null
20.I.3	*	Proband	21							Null
26.I.1	*	Proband	21							Null
390.I.1	*	Proband	22							Null
49.III.2	*	Proband	23							Null
38.I.1	*	Proband	24							Null
203.I.1	*	Proband	26							Null
39.I.1	*	Proband	26							Null
22.I.1	*	Proband	27							Null
340.II.1	*	Proband	30							Null
362.I.1	*	Proband	32							Null
8.I.1	*	Proband	33							Null
401.I.1	*	Proband	33							Null
350.I.1	*	Proband	34							Null
13.II.1	*	Proband	34							Null
233.I.1	*	Proband	34							Null
347.I.1	*	Proband	36							Null
386.II.1	*	Proband	38							Null
357.I.1	*	Proband	39							Null
30.II.1	*	Proband	44							Null
413.I.1	*	Proband	44							Null
411	*	Proband	46							Null
27.I.1	*	Proband	50							Null
129.I.1	*	Proband	53							Null
49.I.1	*	Proband	62							Null
53.V.3	*	Proband	21							Regulatory
342.I.1	*	Proband	30							Regulatory
48.III.14	*	Proband	32							Regulatory
335.III.1	*	Proband	33							Regulatory
6.II.1	*	Proband	36							Regulatory
11.II.1	*	Proband	40							Regulatory
25.II.1	*	Proband	41							Regulatory
4.II.1	*	Proband	54							Regulatory
401.I.2	*	Proband	61							Regulatory
17.II.2	*	Relative	8							Missense
52.I.5	*	Relative	16							Missense
173.I.1	*	Relative	16							Missense
40.II.3	*	Relative	19							Missense
52.II.4	*	Relative	19							Missense
31.II.2	*	Relative	29							Missense
9.III.2	*	Relative	29							Missense
349.III.16	*	Relative	36							Missense
357.I.1	*	Relative	43							Missense
51.II.1	*	Relative	54							Missense
34.I.1	*	Relative	55							Missense
50.II.1	*	Relative	23							Null
39.I.2	*	Relative	28							Null
47.I.1	*	Relative	54							Null
13.II.1	*	Relative	61							Null
4.III.5	*	Relative	15							Regulatory
4.III.4	*	Relative	17							Regulatory
53.V.4	*	Relative	18							Regulatory
4.III.2	*	Relative	29							Regulatory
4.II.2	*	Relative	31							Regulatory
4.II.5	*	Relative	44							Regulatory
48.II.2	*	Relative	53							Regulatory
335.II.1	*	Relative	59							Regulatory
48.II.8	*	Relative	63							Regulatory
25.I.1	*	Relative	66							Regulatory
6.I.1	*	Relative	67							Regulatory
4.I.1	*	Relative	78							Regulatory

Radiographic findings in GATA2 deficiency probands and relatives grouped according to mutation type.

The findings are categorized by the typical patterns seen on chest CT in the GATA2 population described (3 young children in the cohort had no available CT). Finding displayed as present in solid grey or blank for absent. Probands and relatives are grouped separately, according mutation and sorted age. The numbering of the patients follows the scheme in Spinner, Sanchez, et al, Blood 2014

e-Table 3. Pulmonary function tests according to age and status (proband and relatives) FEV1 %, FVC%, and DLCO% adjust for hemoglobin in relatives and probands by decades of life.

Age	<10yrs		10-19yrs		20-29yrs		30-39 yrs.		40-49 yrs.		50-59 yrs.		>60yrs	
	Proband N=3	Proband N=13	Relatives N=7	Proband N=25	Relatives N=4	Proband N=26	Relatives N=3	Proband N=10	Relatives N=2	Proband N=5	Relatives N=4	Proband N=4	Relatives N=3	
FEV1% Median (IQ)	99 (91-107)	85* (78-96)	103 (93-105)	91 (68-100)	90 (66-103)	82 (69-94)	93 (91-106)	71 (60-91)	104 (95-113)	82 (73-92)	88 (85-93)	80 (63-95)	91 (69-108)	
FVC% Median (IQ)	101 (100-103)	99 (84-109)	103 (92-108)	93 (75-105)	97 (89-109)	91 (79--102)	100 (94-113)	89* (67-98)	108 (104-112)	85 (66-91)	95 (89-98)	98 (72-102)	93 (83-94)	
DLCO% adj Median (IQ)	75 (69-82)	71 (58-77)	77 (66-82)	57 (49-74)	71 (57-86)	61* (50-72)	81 (71-93)	66 (54-80)	69 (68-71)	58 (45-83)	79 (68-106)	62 (33-90)	60 (57-77)	

Pulmonary function tests according to age, proband and relatives. FEV1 %, FVC%, and DLCO% adjusted for hemoglobin by decade.

*P value <0.05