Perkins et al 2016

Title: Smith-Magenis syndrome patients often display antibody deficiency but not other immune pathologies

Online Repository Materials:

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Figure Legends

Figure E1: A detailed account of the reactivities of SMS sera versus control sera to 337 antigens. A heat map displays reactivities of sera from 18 SMS subjects 14 healthy unrelated and 3 related controls to the 337 listed antigens. Colorimetric differences on the heat map correspond.

Figure E2: Persistent verrucous disease of the hand (A) and foot (B) of SMSY4

Figure E3: Bivariate analysis of serum reactivity to tetanus toxoid by conventional laboratory testing versus antigen microarray reveals fair concordance. Linearity (R2) and statistical significance (P) calculated via linear regression analysis. MFI, mean florescence intensity.

Table E1- Location, size and relevant genes contained within the deleted regions of 25 SMS blood donors

	deleted element		deleted element encompasses:			
	location	size	*RAI1*	*TOM1L2*	*FLCN*	*TNFRSF13B*
SMS1	17p11.2[a]	1.5 Mb	Y	Y	Y	Y
SMS2	17p11.2[b]	3.6 Mb	Y	Y	Y	Y
SMSY2	17p11.2[b]	3.7 Mb	Y	Y	Y	Y
SMS4	17p11.2[a]	3.4 Mb	Y	Y	Y	Y
SMS5	17p11.2[b]	6.3 Mb	Y	Y	Y	Y
SMSY6	17p11.2[a]	3.4 Mb	Y	Y	Y	Y
SMS7	17p11.2[a]	1.5Mb	Y	Y	Y	Y
SMS8	17p11.2[a]	3.1 Mb	Y	Y	Y	Y
SMS9	17p11.2[b]	1.2 Mb	Y	Y	Y	Y
SMSY3	17p11.2[b]	3.7 Mb	Y	Y	Y	Y
SMS10	17p11.2[b]	3.4 Mb	Y	Y	Y	Y
SMS11	17p11.2[a]	6.4 Mb	Y	Y	Y	Y
SMS12	17p11.2[a]	3.7 Mb	Y	Y	Y	Y
SMS13	17p11.2[b]	3.6 Mb	Y	Y	Y	Y
SMS14	17p11.2[a]	3.6 Mb	Y	Y	Y	Y
SMSY5	17p11.2[a]	6.3 Mb	Y	Y	Y	Y
SMSY7	17p11.2[a]	3.3 Mb	Y	Y	Y	Y
SMS17	17p11.2[a]	3.6 Mb	Y	Y	Y	Y
SMS18	17p11.2[a]	3.2 Mb	Y	Y	Y	Y
SMS19	17p11.2[a]	3.4 Mb	Y	Y	Y	Y
SMS20	17p11.2[a]	3.7 Mb	Y	Y	Y	Y
SMS21	17p11.2[a]	3.4 Mb	Y	Y	Y	Y
SMS22	17p11.2[a]	2.1 Mb	Y	Y	Y	Y
SMSY4	17p11.2[a]	3.5 Mb	Y	Y	Y	Y
SMS23	17p11.2[a]	3.5 Mb	Y	Y	Y	Y

Y, Yes, the indicated gene is deleted

[a] Determined by FISH

[b] Determined by microarray

Table E2- History of autoimmune diseases in 76 SMS subjects (ages 6 months-37 years)

	Percentage (n)
Autoimmune thyroiditis	2.6 (2)
Autoimmune neutropenia	1.3 (1)
Pernicious anemia	1.3 (1)

Table E3- Atopic history in 76 SMS subjects (ages 6 months-37 years)

	Percentage (n)
Allergic Rhinitis	15.8 (12)
Atopic Dermatitis	9.2 (7)
Food Allergy	6.6 (5)
Asthma	7.9 (6)
Drug Allergy	2.6 (2)

Table E4- Relative frequencies of lymphocyte subsets in the peripheral blood of 19 SMS subjects

	Age[a]	Sex	$CD3^+$ (% PBLs)	$CD3^+CD4^+$ (% PBLs)	$CD3^+CD8^+$ (% PBLs)	$CD3^+CD4^+CD25^{hi}$ $FOXP3^+CD127^-$ (% $CD4^+$ T cells)	$CD19^+$ (% PBLs)	$CD19^+CD27^+$ (% B cells)	$CD19^+CD27^+$ IgM^- (% B cells)	$CD3^-$ $CD16^+CD56^+$ (% PBLs)
Reference			-[b]	-[b]	-[b]	-[b]	-[b]	-[b]	-[b]	-[b]
SMS2	6	M	33.5 **(L)**	14.4 **(L)**	18.9	4.6	41.8 **(H)**	13.6	1.2 **(L)**	3.4 **(L)**
SMS4	7	M	56.7	38.3	15.6	2.7 **(L)**	15.5	23.9	2.5 **(L)**	19.1
SMS7	10	M	68.4	28.7	18.0	5.5	19.5	11.8 **(L)**	1.7 **(L)**	4.7
SMS8	10	F	48.0	29.4	17.4	5.4	36.1 **(H)**	41.2 **(H)**	12.0	11.1
SMS9	11	M	63.4	31.1	25.4	5.9	18.6	16.3	3.9 **(L)**	8.1
SMS10	13	F	68.6	28.6	28.0	ND	30.5 **(H)**	9.5 **(L)**	2.8 **(L)**	6.2
SMS11	15	F	54.1	35.9	14.7	4.1	48.0 **(H)**	22.8	4.9 **(L)**	3.6
SMS12	16	F	61.0	37.5	19.1	4.9	26.7	20.1	4.1 **(L)**	4.7
SMS13	16	M	58.9	34.2	21.9	4.5	26.2	29.3	3.5 **(L)**	8.2
SMS14	20	F	63.4	38.8	23.0	5.7	25.3	27.6	7.6 **(L)**	5.2
SMSY5	20	F	56.8	22.2	32.1	6.5[c]	20.1	11.8 **(L)**	1.7 **(L)**[c]	15.7
SMSY7	20	F	52.2	36.5	14.9	4.0[c]	31.6 **(H)**	20.6	3.5 **(L)**[c]	9.4
SMS17	20	F	57.1	33.8	16.8	4.5	33.0 **(H)**	21.6	3.3 **(L)**	5.3
SMS18	21	F	67.9	45.1	19.1	5.4	7.8	5.1 **(L)**	2.6 **(L)**	9.7
SMS19	22	M	69.9	50.3	18.7	4	9.2	20.2	7.0 **(L)**	3.7
SMS20	22	F	65.9	37.4	25.9	4.1	21.5	22.6	4.9 **(L)**	8.6
SMS21	23	M	48.2	22.8	22.1	8.6	39.7 **(H)**	18.1	3.8 **(L)**	11.6
SMSY4	26	F	58.9	33.4	19.6	5.4[c]	20.6	22.5	12.1[c]	5.7
SMS23	27	M	51.1	30.1	19.4	4.8	32.7 **(H)**	24.3	6.5 **(L)**	12.8

F, female; H, higher than normal range; L, below normal range; M, male; ND, not done; PBL, peripheral blood lymphocyte

[a] Age (years) at time of testing

[b] Institutional reference ranges vary by age

[c] Value previously published[3]

Figure E1

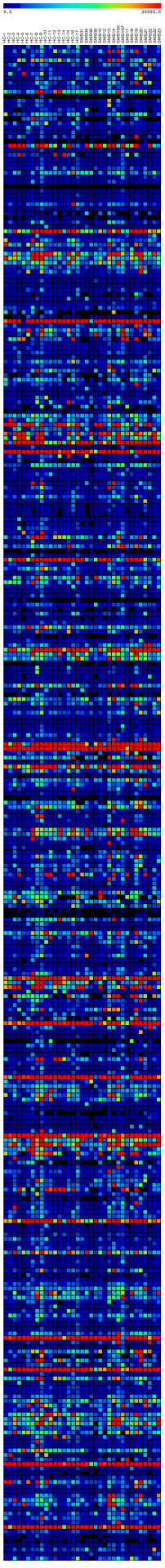

Figure 2E

Figure E3