

Hepatotoxicity_1	SLC22A1	OCT1_G220V	367167	G	G
Hepatotoxicity_1	SLC22A1	OCT1_G38D	366329	G	G
Hepatotoxicity_1	SLC22A1	OCT1_G401S	366084	G	G
Hepatotoxicity_1	SLC22A1	OCT1_G465R	367457	G	G
Hepatotoxicity_1	SLC22A1	OCT1_I403	366304	C	C
Hepatotoxicity_1	SLC22A1	OCT1_L23V	366738	C	C
Hepatotoxicity_1	SLC22A1	OCT1_L85F	366097	C	C
Hepatotoxicity_1	SLC22A1	OCT1_M420del	368437	Ins	Ins
Hepatotoxicity_1	SLC22A1	OCT1_M440I	367166	G	G
Hepatotoxicity_1	SLC22A1	OCT1_N186	366305	C	C
Hepatotoxicity_1	SLC22A1	OCT1_R342H	366085	G	G
Hepatotoxicity_1	SLC22A1	OCT1_R488M	366734	G	G
Hepatotoxicity_1	SLC22A1	OCT1_S14F	366102	C	C
Hepatotoxicity_1	SLC22A1	OCT1_S189L	366092	C	C
Hepatotoxicity_1	SLC22A1	OCT1_S549	366930	A	A
Hepatotoxicity_1	SLC22A1	OCT1_V461I	366735	G	G
Hepatotoxicity_1	SLC22A1	rs1867351	366101	C	C
Hepatotoxicity_1	SLC22A1	rs2282143	366087	C	C
Hepatotoxicity_1	SLC22A1	rs4646277	365795	C	C
Hepatotoxicity_1	SLC22A1	rs4646278	366756	C	C
Hepatotoxicity_1	SLC22A1	rs628031	366500	G	G
Hepatotoxicity_1	SLC22A1	rs683369	366306	C	C
Hepatotoxicity_1	SLC22A2	rs316003	366077	A	A
Hepatotoxicity_1	SLC22A2	rs316019	367040	G	G
Hepatotoxicity_1	SLC22A2	rs3907239	367272	G	G
Hepatotoxicity_1	SLC22A2	rs8177516	366498	C	C
Hepatotoxicity_1	SLC22A2	rs8177517	367539	A	A
Hepatotoxicity_1	SLC22A2	rs8177520	367271	C	C
Hepatotoxicity_1	SLC22A2	SLC22A2_11507C>G	367542	C	C
Hepatotoxicity_1	SLC22A2	SLC22A2_15110C>T	366078	C	C
Hepatotoxicity_1	SLC22A2	SLC22A2_2107T>C	368110	T	T
Hepatotoxicity_1	SLC22A2	SLC22A2_2119A>G	368087	A	A
Hepatotoxicity_1	SLC22A2	SLC22A2_34039C>T	367037	C	C
Hepatotoxicity_1	SLC22A2	SLC22A2_8206C>T	367273	C	C
Hepatotoxicity_1	SLC22A3	rs2292334	366496	C	T
Hepatotoxicity_1	SLC22A3	rs3918291	366497	T	T
Hepatotoxicity_1	SLC22A3	rs8187722	366928	A	A
Hepatotoxicity_1	SLC22A3	SLC22A3_88703C>T	366929	C	C
Hepatotoxicity_1	SLC22A4	rs1050152	367376	C	C
Hepatotoxicity_1	SLC22A4	rs272893	366728	A	G
Hepatotoxicity_1	SLC22A4	rs460415	366729	C	C
Hepatotoxicity_1	SLC22A4	rs4646201	366069	G	G
Hepatotoxicity_1	SLC22A5	rs1045019	367035	G	G
Hepatotoxicity_1	SLC22A5	rs1045020	366727	C	C
Hepatotoxicity_1	SLC22A5	rs2405518	367034	T	T
Hepatotoxicity_1	SLC22A5	rs274548	367684	G	G
Hepatotoxicity_1	SLC22A5	rs274558	366071	C	T
Hepatotoxicity_1	SLC22A6	rs4149170	366063	G	G
Hepatotoxicity_1	SLC22A8	rs2276299	366726	A	T
Hepatotoxicity_1	SLC28A1	rs2242048	366491	C	C
Hepatotoxicity_1	SLC28A1	rs2290272	366292	C	T
Hepatotoxicity_1	SLC28A1	rs3803390	366925	A	G
Hepatotoxicity_1	SLC28A1	rs8042415	367031	A	A
Hepatotoxicity_1	SLC28A1	rs8187737	366495	C	C
Hepatotoxicity_1	SLC28A1	rs8187751	366061	C	C
Hepatotoxicity_1	SLC28A1	rs8187755	366725	A	A
Hepatotoxicity_1	SLC28A1	rs8187758	366326	A	C
Hepatotoxicity_1	SLC28A1	rs8187779	366490	C	C

Hepatotoxicity_1	SLC28A1	SLC28A1_1047C>G	367851	C	C
Hepatotoxicity_1	SLC28A1	SLC28A1_17864C>T	366755	C	C
Hepatotoxicity_1	SLC28A1	SLC28A1_36236A>G	366289	A	A
Hepatotoxicity_1	SLC28A1	SLC28A1_38C>G	366494	C	C
Hepatotoxicity_1	SLC28A1	SLC28A1_45399G>A	366492	G	G
Hepatotoxicity_1	SLC28A1	SLC28A1_47285G>C	366724	G	G
Hepatotoxicity_1	SLC28A1	SLC28A1_55739T>C	366924	T	T
Hepatotoxicity_1	SLC28A1	SLC28A1_57370A>G	366486	A	A
Hepatotoxicity_1	SLC28A1	SLC28A1_57393C>G	366058	C	C
Hepatotoxicity_1	SLC28A1	SLC28A1_57448A>G	366287	A	A
Hepatotoxicity_1	SLC28A2	rs1060896	366923	C	C
Hepatotoxicity_1	SLC28A2	rs2271437	366922	T	T
Hepatotoxicity_1	SLC28A2	rs8023604	366485	T	T
Hepatotoxicity_1	SLC28A3	rs7043492	367029	G	G
Hepatotoxicity_1	SLC28A3	rs7853758	366920	G	G
Hepatotoxicity_1	SLC28A3	rs7867504	367536	C	T
Hepatotoxicity_1	SLC29A1	rs186556	367456	C	C
Hepatotoxicity_1	SLC29A1	rs8187630	366720	G	G
Hepatotoxicity_1	SLC29A1	SLC29A1_10817G>A	366325	G	G
Hepatotoxicity_1	SLC29A1	SLC29A1_10867T>C	367268	T	T
Hepatotoxicity_1	SLC29A1	SLC29A1_10907G>A	366483	G	G
Hepatotoxicity_1	SLC29A1	SLC29A1_12610T>C	367535	T	T
Hepatotoxicity_1	SLC29A1	SLC29A1_13816C>A	366055	C	C
Hepatotoxicity_1	SLC29A2	rs8187661	366050	C	C
Hepatotoxicity_1	SLC29A2	SLC29A2_2125G>A	366285	G	G
Hepatotoxicity_1	SLC29A2	SLC29A2_2152C>A	366717	C	C
Hepatotoxicity_1	SLC29A2	SLC29A2_2405G>A	365790	G	G
Hepatotoxicity_1	SLC29A2	SLC29A2_3724delT	368156	Ins	Ins
Hepatotoxicity_1	SLC29A2	SLC29A2_3725delG	368158	Ins	Ins
Hepatotoxicity_1	SLC29A2	SLC29A2_3726delG	368162	Ins	Ins
Hepatotoxicity_1	SLC29A2	SLC29A2_7341G>A	366716	G	G
Hepatotoxicity_1	SLC5A6	rs1064845	366480	G	G
Hepatotoxicity_1	SLC5A6	rs1275524	366283	G	G
Hepatotoxicity_1	SLC5A6	rs1395	366048	A	A
Hepatotoxicity_1	SLC5A6	rs4327199	366715	G	G
Hepatotoxicity_1	SLC5A6	rs7081	366282	A	A
Hepatotoxicity_1	SLC7A5	rs1060250	367265	C	C
Hepatotoxicity_1	SLC7A5	rs1060253	366919	C	G
Hepatotoxicity_1	SLC7A5	rs2287119	368130	T	T
Hepatotoxicity_1	SLC7A7	rs1061040	367164	G	G
Hepatotoxicity_1	SLC7A7	rs1805059	366708	A	A
Hepatotoxicity_1	SLC7A7	rs1805061	367165	C	T
Hepatotoxicity_1	SLC7A7	rs2281677	366280	C	C
Hepatotoxicity_1	SLC7A7	rs8016634	366043	G	G
Hepatotoxicity_1	SLC7A7	rs8018462	366042	G	G
Hepatotoxicity_1	SLC01A2	rs4078	367548	A	A
Hepatotoxicity_1	SLC01A2	rs7298982	367549	T	T
Hepatotoxicity_1	SLC01A2	rs7312628	367465	C	C
Hepatotoxicity_1	SLC01A2	rs7484455	367384	G	G
Hepatotoxicity_1	SLC01A2	rs7957203	367637	A	T
Hepatotoxicity_1	SLC01B1	OATPC_A1294G	367636	A	A
Hepatotoxicity_1	SLC01B1	OATPC_A1385G	367377	A	A
Hepatotoxicity_1	SLC01B1	OATPC_A2000G	367632	A	A
Hepatotoxicity_1	SLC01B1	OATPC_A467G	367379	A	A
Hepatotoxicity_1	SLC01B1	OATPC_C463A	367382	C	C
Hepatotoxicity_1	SLC01B1	OATPC_T245C	367588	T	T
Hepatotoxicity_1	SLC01B1	OATPCstar10_A1964G	367635	A	A
Hepatotoxicity_1	SLC01B1	OATPCstar2_T217C	367688	T	T

Hepatotoxicity_1	SLCO1B1	OATPCstar6_T1058C	367463	T	T
Hepatotoxicity_1	SLCO1B1	OATPCstar9_G1463C	366932	G	G
Hepatotoxicity_1	SLCO1B1	rs2291075	367471	C	T
Hepatotoxicity_1	SLCO1B1	rs2306283	367686	A	G
Hepatotoxicity_1	SLCO1B1	rs4149056	367545	T	T
Hepatotoxicity_1	SLCO1B1	rs4149057	367170	C	T
Hepatotoxicity_1	SLCO1B3	rs2053098	367042	A	G
Hepatotoxicity_1	SLCO1B3	rs3764006	367041	T	T
Hepatotoxicity_1	SLCO1B3	rs4149117	367543	G	T
Hepatotoxicity_1	SLCO1B3	rs4149143	367460	A	A
Hepatotoxicity_1	SLCO1B3	rs7311358	367461	A	G
Hepatotoxicity_1	SLCO2B1	rs1621378	366308	C	C
Hepatotoxicity_1	SPG7	rs12960	366040	A	G
Hepatotoxicity_1	SPG7	rs2099104	366475	T	T
Hepatotoxicity_1	SPG7	rs2292954	366279	C	T
Hepatotoxicity_1	SULT1A1	rs1042005	367899	G	G
Hepatotoxicity_1	SULT1A1	rs1042008	367850	C	C
Hepatotoxicity_1	SULT1A1	rs1042011	367770	G	G
Hepatotoxicity_1	SULT1A1	rs1042014	367786	G	G
Hepatotoxicity_1	SULT1A1	rs1042157	367237	C	C
Hepatotoxicity_1	SULT1A1	rs4987024	366649	A	A
Hepatotoxicity_1	SULT1A3	rs1059667	367705	T	T
Hepatotoxicity_1	SULT1B1	rs4149416	367654	A	A
Hepatotoxicity_1	SULT1C1	rs1047309	367620	A	A
Hepatotoxicity_1	SULT1C1	rs1047310	367389	G	G
Hepatotoxicity_1	SULT1C1	rs14700	367653	A	A
Hepatotoxicity_1	SULT1C1	rs3731680	367122	T	T
Hepatotoxicity_1	SULT1C2	rs1402467	367353	C	C
Hepatotoxicity_1	SULT1E1	rs3736599	367568	A	G
Hepatotoxicity_1	SULT2B1	rs1052124	366227	C	C
Hepatotoxicity_1	SULT2B1	rs1132054	366226	C	C
Hepatotoxicity_1	SULT2B1	rs2302947	365966	C	C
Hepatotoxicity_1	SULT2B1	rs2544794	365964	G	G
Hepatotoxicity_1	SULT4A1	rs138056	366437	A	C
Hepatotoxicity_1	SULT4A1	rs138057	366648	A	G
Hepatotoxicity_1	SULT4A1	rs3179394	366873	G	G
Hepatotoxicity_1	TBXAS1	CYP5A1star1B	366409	G	G
Hepatotoxicity_1	TBXAS1	CYP5A1star1D	367335	G	G
Hepatotoxicity_1	TBXAS1	CYP5A1star2	367220	G	G
Hepatotoxicity_1	TBXAS1	CYP5A1star3	366619	C	C
Hepatotoxicity_1	TBXAS1	CYP5A1star4	366407	A	A
Hepatotoxicity_1	TBXAS1	CYP5A1star5	366618	C	C
Hepatotoxicity_1	TBXAS1	CYP5A1star7	366617	NC	NC
Hepatotoxicity_1	TBXAS1	CYP5A1star8	366615	C	C
Hepatotoxicity_1	TBXAS1	rs1042561	365934	C	C
Hepatotoxicity_1	TBXAS1	rs3735354	365931	G	G
Hepatotoxicity_1	TBXAS1	rs4526	367803	G	G
Hepatotoxicity_1	TBXAS1	rs4527	365929	C	C
Hepatotoxicity_1	TBXAS1	rs5760	366211	G	G
Hepatotoxicity_1	TBXAS1	rs5761	365930	G	G
Hepatotoxicity_1	TBXAS1	rs5762	367817	C	C
Hepatotoxicity_1	TBXAS1	rs5769	367505	A	A
Hepatotoxicity_1	TBXAS1	rs5770	367278	A	A
Hepatotoxicity_1	TBXAS1	rs5771	365933	C	C
Hepatotoxicity_1	TBXAS1	rs6137	366613	C	C
Hepatotoxicity_1	TBXAS1	rs6140	366612	T	T
Hepatotoxicity_1	TBXAS1	rs8192833	366986	G	G
Hepatotoxicity_1	TBXAS1	rs8192864	365926	G	G

Hepatotoxicity_1	TPMT	rs1142345	367679	A	A
Hepatotoxicity_1	TPMT	rs1800462	367234	G	G
Hepatotoxicity_1	TPMT	rs1800584	367652	G	G
Hepatotoxicity_1	TPMT	rs2842934	366871	T	T
Hepatotoxicity_1	TPMT	rs2842944	366872	C	C
Hepatotoxicity_1	TPMT	rs6921269	367235	G	G
Hepatotoxicity_1	TPMT	TPMTstar3B	367281	G	G
Hepatotoxicity_1	TPMT	TPMTstar8	368264	G	G
Hepatotoxicity_1	UGT1A1	UGT1A1_T247C	366641	T	T
Hepatotoxicity_1	UGT1A1	UGT1A1_T-3279G	367228	T	T
Hepatotoxicity_1	UGT1A1	UGT1A1star1B_1813CT	368422	C	C
Hepatotoxicity_1	UGT1A1	UGT1A1star1B_1941CG	368425	C	C
Hepatotoxicity_1	UGT1A1	UGT1A1star1B_2042CG	368427	C	C
Hepatotoxicity_1	UGT1A1	UGT1A1star27	366435	C	C
Hepatotoxicity_1	UGT1A1	UGT1A1star28	1684360	Ins	Del
Hepatotoxicity_1	UGT1A1	UGT1A1star28-3156GA	368419	G	G
Hepatotoxicity_1	UGT1A1	UGT1A1star29	366433	C	C
Hepatotoxicity_1	UGT1A1	UGT1A1star43	366642	T	T
Hepatotoxicity_1	UGT1A1	UGT1A1star6	367229	G	G
Hepatotoxicity_1	UGT1A1	UGT1A1star7	366645	T	T
Hepatotoxicity_1	UGT1A8	rs1126788	368109	T	T
Hepatotoxicity_1	UGT1A8	rs1126790	368118	G	G
Hepatotoxicity_1	UGT1A8	rs1126792	368119	T	T
Hepatotoxicity_1	UGT1A8	rs1126793	368127	C	C
Hepatotoxicity_1	UGT1A8	rs1126798	367513	A	A
Hepatotoxicity_1	UGT1A8	rs1126804	367440	A	A
Hepatotoxicity_1	UGT1A9	rs1042707	366999	A	A
Hepatotoxicity_1	UGT1A9	rs1042708	367724	C	C
Hepatotoxicity_1	UGT1A9	rs1105880	366225	A	A
Hepatotoxicity_1	UGT1A9	rs1126803	368061	A	A
Hepatotoxicity_1	UGT1A9	rs2011404	366222	C	C
Hepatotoxicity_1	UGT1A9	rs4148323	367227	G	G
Hepatotoxicity_1	UGT1A9	rs6431625	365957	C	T
Hepatotoxicity_1	UGT1A9	rs6759892	366863	T	T
Hepatotoxicity_1	UGT1A9	rs7577677	366870	C	C
Hepatotoxicity_1	UGT2A1	rs1347046	367340	A	A
Hepatotoxicity_1	UGT2A1	rs4148301	367511	G	G
Hepatotoxicity_1	UGT2A1	rs4148304	366861	G	G
Hepatotoxicity_1	UGT2A1	rs4148310	367566	A	A
Hepatotoxicity_1	UGT2A1	rs4148311	367512	G	G
Hepatotoxicity_1	UGT2B11	rs3890590	367439	G	G
Hepatotoxicity_1	UGT2B11	rs7697037	366428	A	G
Hepatotoxicity_1	UGT2B15	rs1902023	367703	G	G
Hepatotoxicity_1	UGT2B4	rs1051752	367678	A	A
Hepatotoxicity_1	UGT2B4	rs1845555	366996	G	G
Hepatotoxicity_1	UGT2B4	rs1966151	367723	A	G
Hepatotoxicity_1	UGT8	rs4148255	366640	A	A
Hepatotoxicity_1	UGT8	rs6851610	367048	G	G
Hepatotoxicity_1	VKORC1	rs1046030	1684406	C	C
Hepatotoxicity_1	VKORC1	rs11540137	1684395	C	C
Hepatotoxicity_1	VKORC1	rs13338824	1684413	A	A
Hepatotoxicity_1	VKORC1	rs17708472	1684409	G	G
Hepatotoxicity_1	VKORC1	rs17878544	1684407	A	A
Hepatotoxicity_1	VKORC1	rs17880887	1684394	C	C
Hepatotoxicity_1	VKORC1	rs17884388	1684414	T	T
Hepatotoxicity_1	VKORC1	rs17884850	1684412	G	G
Hepatotoxicity_1	VKORC1	rs17884982	1684403	A	A
Hepatotoxicity_1	VKORC1	rs17886199	1684411	T	T

Hepatotoxicity_1	VKORC1	rs1895514	1684398	T	T
Hepatotoxicity_1	VKORC1	rs2359612	1684432	T	T
Hepatotoxicity_1	VKORC1	rs2884737	1684418	T	T
Hepatotoxicity_1	VKORC1	rs7196161	1684445	T	T
Hepatotoxicity_1	VKORC1	rs7200749	1684422	C	C
Hepatotoxicity_1	VKORC1	rs8050894	1684431	C	C
Hepatotoxicity_1	VKORC1	rs9923231	1684447	A	A
Hepatotoxicity_1	VKORC1	rs9934438	1684424	A	A
Hepatotoxicity_1	VKORC1	VKORC1_A172G	1684429	A	A
Hepatotoxicity_1	VKORC1	VKORC1_C292T	1684393	C	C
Hepatotoxicity_1	VKORC1	VKORC1_G196A	1684392	G	G
Hepatotoxicity_1	VKORC1	VKORC1_T383G	1684438	T	T
Hepatotoxicity_1	XDH	rs1042036	367114	C	C
Hepatotoxicity_1	XDH	rs1884725	367339	G	G
Hepatotoxicity_1	XDH	rs207440	365954	G	G
Hepatotoxicity_1	XDH	rs2295474	367113	C	C
Hepatotoxicity_1	XDH	rs2295475	366639	A	A
Hepatotoxicity_1	XDH	rs4407290	366995	G	G
Hepatotoxicity_1	XDH	rs566362	366638	G	G
Hepatotoxicity_1	XDH	rs669884	366425	G	G

添付資料8: DILI患者検体12検体のDMET 2.0 解析結果

gene	polymorphism	Control (n=60)			Hepatotoxicity (n=24)		
		Wild allele	Variant allele	Frequency (variant)	Wild allele	Variant allele	Frequency (variant)
ABCB1	3435C>T	36	24	0.40	14	10	0.42
CYP2B6	CYP2B6*2B	58	2	0.03	23	1	0.04
CYP2B6	CYP2B6*3	60	0	0.00	24	0	0.00
CYP2C19	CYP2C19*2	37	23	0.38	15	9	0.37
CYP2C19	CYP2C19*3	49	11	0.18	22	2	0.08
CYP2C9	CYP2C9*2	60	0	0.00	24	0	0.00
CYP2C9	CYP2C9*3	60	0	0.00	24	0	0.00
CYP2D6	CYP2D6*4	60	0	0.00	24	0	0.00
CYP2D6	CYP2D6*10	33	27	0.45	17	7	0.29
CYP3A5	CYP3A5*3	11	49	0.82	6	18	0.75
CYP3A5	CYP3A5*6	60	0	0.00	24	0	0.00
DPYD	DPDY*2A	60	0	0.00	24	0	0.00
DPYD	DPDY*3	58	0	0.00	24	0	0.00
TPMT	TPMT*3C	60	0	1.00	24	0	0.00
UGT1A1	UGT1A1*6	51	9	0.15	20	4	0.17
UGT1A1	UGT1A1*28	54	6	0.10	22	2	0.08

○本表はallele数を基準に記載したため、n数は2×検体数となる。

○Controlには、独立した臨床研究において、薬物動態関連遺伝子を含む遺伝子の解析に対する同意取得済みのDNA検体を連結不可能匿名化の上利用した。

○Allele頻度はvariant alleleの頻度を示す。

○添付資料7に示したSNPsの中から、機能的影響が大きい代表的なSNPsを選択した。

○DPDY*3のみコントロール1検体で測定不能であったため、allele数は58となっている。