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原　寿郎	II　中枢神経系感染症　その他の中枢神経系感染症急性散在性脳脊髄炎（ADEM）	別冊日本臨牀	25	144-7	2013
原　寿郎	＜Clinical Science＞自然免疫が関与する炎症性疾患：川崎病	炎症と免疫	21	62-7	2013
原　寿郎	こどもの発熱の原因とその対処法	ふたば	77		2013
戸田尚子、原　寿郎	2.疾患と栄養　先天性免疫不全症と低栄養	臨床栄養		印刷中	2014
有賀　正	補体欠損症	小児科診療	76	453-8	2013
有賀　正	乳児一過性低ガンマグロブリン血症　液症候群II	日本臨床	22	223-6	2013
有賀　正	Bruton型無ガンマグロブリン血症　血液症候群II	日本臨床	22	227-31	2013
有賀　正	IgG3欠損症　血液症候群III	日本臨床	23	679-81	2013
山崎康博、有賀　正	扁桃がみえない	小児内科	45	1876-8	2013
今井耕輔	原発性免疫不全症の最新国際分類(2011)	小児科	54	1491-515	2013
今井耕輔	原発性免疫不全症における遺伝学的検査の取り扱いと諸問題について	日本小児血液・がん学会雑誌	50	415-7	2013
今井耕輔	自然免疫について. チャイルドヘルス	チャイルドヘルス	16	608-13	2013
内尾寛子、額田貴之、井庭憲人、深尾大輔、橋本有紀子、田部有香、井上美保子、濱畑啓悟、吉田 晃、百井亨、河合朋樹、西小森隆太、平家俊男	NEMO蛋白異常をフローサイトメトリーにより早期診断した色素失調症の新生児例	日本小児科学会雑誌	117	1303-7	2013
河合朋樹、平家俊男	メンデル遺伝型マイコバクテリア感染症	臨床免疫・アレルギー科	60	548-52	2013
阿部純也、西小森隆太、平家俊男	Aicardi-Goutieres症候群	アレルギー・免疫	20	62-9	2013
河合朋樹、平家俊男	患者レジストリーと遺伝子診断	アレルギー・免疫	20	14-23	2013
中村和洋, 小林正夫	新生児同種免疫性好中球減少症	臨床免疫・アレルギー科	60	78-82	2013
波多野修一、駒澤克孝、西村真一郎、藤江篤志、大野令央義、川口 浩史、小林正夫、高尾信一	マイコプラズマ感染症検査法の検討　マイコプラズマ抗原迅速診断キットの有用性について	小児科臨床	66	2105-2115	2013
下村 麻衣子, 千々松 郁枝, 浅野 孝基, 古江 綾, 三木 瑞香, 川口 浩史, 中村和洋, 小林 正夫	慢性肉芽腫症における消化管病変	広島医学	66	473-474	2013
高坂 卓馬, 秀 道広, 小林 正夫	Omenn症候群の1例	西日本皮膚科	75	269	2013
唐川修平、中村和洋、小林正夫	【クローズアップ　新しい子どもの病気】 血液腫瘍疾患　新しい診断技術で診断可能となった疾患　好中球減少症　遺伝子変異と抗好中球抗体.	小児内科	45	1131-3	2013
三木瑞香、小林正夫	【クローズアップ　負荷試験の実際2013】 血液系機能検査　好中球減少症の負荷試験	小児内科	45	989-91	2013
平田　修、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】 リンパ球の異常　リンパ球機能異常と類縁疾患　原発性免疫不全症候群　単独IgGサブクラス欠損症	日本臨床別冊血液症候群第2版II		250-3	2013
早川誠一、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】 リンパ球の異常　リンパ球機能異常と類縁疾患　原発性免疫不全症候群　IgM単独(選択的)欠損症	日本臨床別冊血液症候群第2版II		246-9	2013
平田　修、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】 リンパ球の異常　リンパ球機能異常と類縁疾患　原発性免疫不全症候群　選択的IgA欠損症	日本臨床別冊血液症候群第2版II		242-5	2013

平田　修、中村和洋、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く)　好中球の異常　好中球減少症　周期性好中球減少症	日本臨床別冊血液症候群第2版II		57-60	2013
唐川修平、中村和洋、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く)　好中球の異常　好中球減少症　自己免疫性好中球減少症	日本臨床別冊血液症候群第2版II		54-6	2013
溝口洋子、中村和洋、小林正夫	【血液症候群(第2版)-その他の血液疾患を含めて-】白血球(顆粒球)の異常(悪性腫瘍を除く)　好中球の異常　好中球減少症　同種免疫性好中球減少症.	日本臨床別冊血液症候群第2版II		50-3	2013
唐川修平、小林正夫	【知っておきたい最新の免疫不全症分類-診断から治療まで】 治療　原発性免疫不全症に対する造血幹細胞移植	小児科診療	76	476-80	2013
宮地隆史、丸山博文、小林正夫、松本昌泰	【クローズアップ　呼吸管理】 <在宅呼吸ケア> 在宅人工呼吸器装着者の災害時対策	小児内科	45	116-20	2013
布井博幸	過剰炎症と免疫療法	小児科診療	76	467-75	2013
布井博幸	慢性肉芽腫症の病態解析	血管炎　増刊号		601-4	2013
布井博幸、西村豊樹、中原彰彦	インフルエンザ感染における重症合併症（インフルエンザ脳症）	神経感染症　第2版		652-6	2013
布井博幸	Jordans異常	血液症候群　第2版		101-4	2013
斎藤潤、中畑龍俊	疾患特異的iPS細胞	再生医療	12	19-29	2013
中畑龍俊、岡野光夫、高橋政代	再生医療の現状と将来	HUMAN SCIENCE	24	4-13	2013
中畑龍俊	総論 疾患iPS細胞の樹立と臨床病態解析への応用	Medical Science Digest(MSD)	39	4-6	2013
峯岸克行	STAT3の異常によるアトピー性皮膚炎の発症機序	臨床・免疫アレルギー科	59	160-4	2013
峯岸克行	高IgE症候群の最近の知見	Medical Science Digest	39	7-8	2013
峯岸克行	抗体産生不全症—B細胞不全症	小児科診療	76	419-23	2013
峯岸克行	Jak-Statシグナルとアレルギー制御	実験医学	31	113-7	2013
峯岸克行	高IgE症候群に見られる易感染性	化学療法の領域	29	2429-34	2013
峯岸克行	高IgE症候群	小児内科	45	1146-7	2013
金兼弘和、宮脇利男	免疫担当細胞機能異常検査（リンパ球）	小児科診療	76	180-5	2013
星野顕宏、金兼弘和	X-linked lymphoproliferative syndrome(XLP)/ signaling-lympho-cytic-activation-molecule-associated protein (SAP) の病態	血液フロンティア	23	33-40	2013
笹原洋二	免疫不全症を伴う特徴的な症候群　—Wiskott-Aldrich症候群、高IgE症候群	小児科診療	76	407-12	2012
河野　肇	自然免疫と動脈硬化との関連が指し示す適応とその破綻	適応医学	17	13	2012
河野　肇	動脈硬化の発症メカニズム:免疫反応のかかわり	医学のあゆみ	245	1107-201	2012
原　寿郎	原発性免疫不全症(PID)－悪性腫瘍，血液疾患を合併するPIDを中心に－	日本小児血液・がん学会雑誌	49(3)	237-41	2012
高田英俊、大賀正一、原　寿郎	自己炎症症候群 特集　知っておきたい最新の免疫不全症候群分類－診断から治療まで－	小児科診療	76(3)		2012
星野顕宏、金兼弘和、西田直徳、野村恵子、大賀正一、宮脇利男	抗菌薬投与により治癒し得た再発性肝膿瘍を合併した慢性肉芽腫症の一例	小児感染免疫	24	175-9	2012
有賀　正	小児疾患の診断治療基準。Wiskott-Aldrich症候群。	小児内科	44 (増刊号)	230-1	
有賀　正	知っておきたい内科症候群:重症複合免疫不全症(Omenn症候群を含む)	臨床雑誌　内科	増大号 109	1499-500	2012
大倉有加、有賀　正	アデノシンデアミナーゼ欠損症・アデノシンデアミナーゼ部分欠損症	日本臨床(新領域別症候群シリーズ19「先天性代謝異常症候群」第2版)		571-4	2012
山田雅文、有賀　正	慢性皮膚粘膜カンジダ症と自然免疫異常	Bio Clinica		印刷中	2012

大倉有加、山田雅文、小林一郎、Santisteban I、Arredondo-Santisteban G、加藤善一郎、井口晶裕1、吉田真、小原収、中川紀子、今井耕輔、Hershfield MS、有賀 正	Best articles of the year. ADA酵素蛋白の安定性を相乗的に阻害す"WAZA-ARI" 変異を認めたADA欠損症の1 例。	北海道医学雑誌	87	4-5	2012
有賀 正	総論:遺伝子治療	小児科診療	5	107-14	2012
森尾友宏	先天性免疫不全症の病態と思春期以降のマネジメント	血液内科	65	599-607	2012
森尾友宏	【クローズアップ感染症】<感染性疾患の基礎的な知見の進歩・概念の変化>感染症と自然免疫	小児内科	44	959-65	2012
森尾友宏	【サイトカインのすべて(完全改訂版)】サイトカイン投与およびサイトカイン抑制による治療 免疫不全症	臨床免疫・アレルギー科	57	838-44	2012
森尾友宏	原発性免疫不全症における臨床遺伝学 T細胞系免疫異常症における遺伝子診療	日本遺伝カウンセリング	33	49-53	2012
森尾友宏	分類不能型免疫不全症	日本臨牀	70	2011-21	2012
今井耕輔	原発性免疫不全症の最新国際分類	臨床免疫・アレルギー科	58	446-66	2012
今井耕輔	原発性免疫不全症の遺伝子診断・治療	臨床血液	53	1865-73	2012
今井耕輔	特集 乳幼児健診Q&A Ⅶ.歯科 Q 口の中にミルクのかすのようなものがいつもあるのですが、大丈夫ですか	小児科診療	11	2045-8	2012
今井耕輔	臨床検査の意義と限界 T細胞・B細胞サブセット	小児内科	44	645-8	2012
今井耕輔	【知っておきたい内科症候群】膠原病・免疫・アレルギー《免疫不全症》 ウィスコット・アルドリッチ症候群	内科	109	1501-3	2012
今井耕輔	高IgM	小児内科	44	228-9	2012
近藤直実	毛細血管拡張性小脳失調症	小児疾患の診断治療基準 第4版. 小児内科	44	232-3	2012
大西秀典、寺本貴英、久保田一生、山本崇裕、渡邊倫子、加藤善一郎、金子英雄、近藤直実	原発性(先天性)免疫不全症の分類－2012update－	小児科診療	75	134-51	2012
谷内江昭宏	原発性免疫不全症と体細胞キメリズム	感染・炎症・免疫	42	61-4	2012
西小森隆太、井澤和司、平家俊男	【クローズアップ感染症】PFAPA以外の周期性発熱症候群についての知見(解説/特集)	小児内科	44(7)	1221-6	2012
西小森隆太、井澤和司、平家俊男	原発性免疫不全症における臨床遺伝学 日本における自己炎症疾患の遺伝子診断について(解説)	日本遺伝カウンセリング学会誌	33(1)	63-8	2012
西小森隆太、平家俊男	自己炎症症候群	小児内科	44増刊号	290-1	2012
溝口洋子、鎌田綾、三木瑞香、谷博雄、世羅康彦、中村和洋、小林正夫	Glanzmann thrombasthenia への遺伝子組み換え活性型第VII因子製剤による止血効果	日本小児血液・がん学会雑誌	49(1-2)9	61-6	2012
梶梅輝之、浅野孝基、世羅康彦、小林正夫	輸血後蕁麻疹発症前の末梢血一般検査所見	アレルギー	61(8)	1086-91	2012
溝口洋子、津村弥来、岡田賢、小林正夫	慢性性皮膚粘膜カンジダ症と機能獲得性STAT1変異	臨床免疫・アレルギー科	57(4)	437-43	2012
横田俊平、西小森隆太、高田英俊、菊地雅子、野澤智、金高太一、木澤敏毅、宮前多佳子、森雅亮、平家俊男、原寿郎、今川智之	クリオピリン関連周期性発熱症候群に対する生物学的製剤治療の手引き(2012) カナキヌマブ(総説)	日本小児科学会雑誌	116(9)	1337-41	2012
横田俊平、菊池雅子、野澤智、木澤敏毅、金高太一、宮前多佳子、森雅亮、西小森隆太、高田英俊、平家俊男、原寿郎、今川智之	クリオピリン関連周期性発熱症候群に対するヒト化抗IL-1・モノクローナル抗体製剤カナキヌマブの効果	日本臨床免疫学会雑誌	35	23-9	2013
中畑龍俊	白血病治療の進歩と今後の展望	日本小児血液・がん学会雑誌	49	11-5	2012
中畑龍俊、丹羽明	幹細胞増幅(第10章 内科疾患と再生医療)	カラー版内科学(総集編)		447-50	2012
斎藤潤、中畑龍俊	疾患特異的iPS細胞	再生医療	12	19-32	2012
峯岸克行	高IgE症候群.	知っておきたい内科症候群	109	1495-6	2012
峯岸克行	高IgM症候群	知っておきたい内科症候群	109	1497-8	2012
峯岸克行	高IgE症候群と感染症	化学療法の領域	27	80-4	2012
峯岸克行	高IgE症候群の原因遺伝子解析の現況と臨床応用の可能性	日本医事新報	4610	57-8	2012

峯岸克行	高IgE症候群により発症するアトピー性皮膚炎	臨床・免疫アレルギー科	58	667-70	2012
河野　肇	動脈硬化と自然免疫	Annual Review 循環器 2012		42-8	2012
河野　肇	コレステロール、動脈硬化、そして自然免疫炎症	自己抗体と自己免疫 2012		33-41	2012
河野　肇	NETs(好中球細胞外traps)はSLEの病態に関与するか？	リウマチ科	48	569-73	2011
河野　肇	NLRP3インフラマソーと動脈硬化	臨床免疫・アレルギー科	57	444-9	2011
河野　肇	インフラマソームと動脈硬化	Medical Practice	29	1206-8	2011
原　寿郎	綜説シリーズ—現代医学の焦点(347) 原因不明の発熱と自己炎症性疾患	日本臨牀	69(9)	1679 -89	2011
原　寿郎	病態にせまる（研究成果から本態にせまる） サイトカインネットワークから	小児科診療	74(8)	1114-9	2011
金兼弘和、大坪慶輔、宮脇利男	制御性T細胞に異常を有する原発性免疫不全症	炎症と免疫	19	210-6	2012
大坪慶輔、金兼弘和、宮脇利男	アレルギー疾患と免疫調節（Treg細胞）	小児科	52	879-87	2011
金兼弘和、清水篤実、渡邉健一郎、種市尋宙、宮脇利男	Shwachman-Diamond症候群	日本臨床別冊 新領域別症候群シリーズ	16	65-8	2011
有賀　正	総論：遺伝子治療	小児科診療	75	107-14	2011
右田清志、上松一永	家族性地中海熱の臨床	日本臨床免疫学会誌	34	335-60	2011
谷内江昭宏	EBV-HLHの病態　早期診断・早期治療介入のためのサイトカインプロファイリングと細胞解析	小児感染免疫	23	43-50	2011
酒井秀政、平家俊男	日本における高IgD症候群の診断と展望	日本臨床免疫学会雑誌	34(5)	382-7	2011
中畑龍俊	疾患特異的iPS細胞を用いた遺伝子治療・個別化医療	小児科	52	1743-9	2011
土屋　滋	総説　原発性免疫不全症に対する造血幹細胞移植	小児感染免疫	22(4)	381-4	2011
笹原洋二	総説　Wiskott-Aldrich症候群の分子病態からみた感染症とWIPの役割	小児感染免疫	23(1)	75-80	2011
峯岸克行	原発性免疫不全症の新展開	炎症と免疫	19(5)	3-4	2011
峯岸克行	高IgE症候群における易感染性のメカニズム	呼吸器内科	20	83-6	2011
齋藤雅子、峯岸克行	高IgE症候群の病因・病態解析	アレルギー・免疫	18	72-6	
河野　肇	NLRP3と痛風	高尿酸血症と痛風	19	59-65	
河野　肇	NLRP3インフラマソームと動脈硬化	呼吸と循環	59	873-82	

Ⅲ　研究成果の刊行物

nature genetics

Exome sequencing identifies secondary mutations of *SETBP1* and *JAK3* in juvenile myelomonocytic leukemia

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Juvenile myelomonocytic leukemia (JMML) is an intractable pediatric leukemia with poor prognosis[1] whose molecular pathogenesis is poorly understood, except for somatic or germline mutations of RAS pathway genes, including *PTPN11*, *NF1*, *NRAS*, *KRAS* and *CBL*, in the majority of cases[2–4]. To obtain a complete registry of gene mutations in JMML, whole-exome sequencing was performed for paired tumor-normal DNA from 13 individuals with JMML (cases), which was followed by deep sequencing of 8 target genes in 92 tumor samples. JMML was characterized by a paucity of gene mutations (0.85 non-silent mutations per sample) with somatic or germline RAS pathway involvement in 82 cases (89%). The *SETBP1* and *JAK3* genes were among common targets for secondary mutations. Mutations in the latter were often subclonal and may be involved in the progression rather than the initiation of leukemia, and these mutations associated with poor clinical outcome. Our findings provide new insights into the pathogenesis and progression of JMML.

JMML is a rare myelodysplastic/myeloproliferative neoplasm unique to childhood, characterized by excessive proliferation of myelomonocytic cells and hypersensitivity to granulocyte-macrophage colony-stimulating factor[1]. A cardinal genetic feature of JMML is frequent somatic and/or germline mutation of RAS pathway genes, such as *NF1*, *NRAS*, *KRAS*, *PTPN11* and *CBL*, which are mutated in more than 70% of JMML cases in a mutually exclusive manner[2–4]. However, it is still open to question whether RAS pathway mutations are sufficient for the development of JMML or if secondary mutations have a role in the development and progression of this cancer. To address these issues and to better define the molecular pathogenesis of JMML, we performed whole-exome sequencing of paired tumor-normal DNA from 13 cases (**Supplementary Table 1**). We obtained mean coverage in exome sequencing of 137× for tumor samples and 143× for normal samples (**Supplementary Fig. 1**). A Monte-Carlo simulation indicated that the study detected 88% of the existing somatic mutations (Online Methods and **Supplementary Fig. 2**).

Sanger sequencing of 25 candidate non-silent somatic nucleotide alterations confirmed 1 nonsense and 10 missense mutations (**Table 1** and **Supplementary Fig. 3**), with the low true positive rate consistent with the very low numbers of somatic mutations in JMML. Of the 11 somatic mutations, 6 involved known RAS pathway genes. In addition, non-overlapping RAS pathway mutations (6 somatic and 6 germline) were confirmed in 11 of the 13 discovery cases (86%; **Table 1**). For the remaining two cases that lacked documented RAS pathway mutations, we intensively searched for possible germline mutations that could be relevant to the development of JMML. In total, 179 and 167 candidate germline mutations were detected in subjects 77 and 92, respectively, but these mutations did not affect known RAS pathway genes or other cancer-related genes, including the ones registered in the pathway databases (Online Methods). A frameshift deletion in *KMT2D* (also known as *MLL2*; encoding p.Val1670fs) was found in subject 92, who had been diagnosed as having Noonan syndrome on the basis of typical features such as hypertelorism, webbed neck and congenital heart disease (**Supplementary Fig. 3**) but lacked the distinctive facial appearance of Kabuki syndrome, which was shown to be caused by germline *KMT2D* mutations[5].

Five of the 11 somatic mutations were non–RAS pathway mutations, involving *SETBP1* (3 p.Asp868Asn alterations), *JAK3* (1 p.Arg657Gln alteration) and *SH3BP1* (1 p.Ser277Leu alteration), which had not been reported in JMML cases. SETBP1 was originally isolated as a 170-kDa nuclear protein that interacts with SET, a small protein inhibitor of the putative tumor suppressors PP2A and NM23-H1 (ref. 6). Several lines of recent evidence suggest that SETBP1 has a role in leukemogenesis (**Supplementary Fig. 4**)[7–11]. *SETBP1* participates in

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Table 1 List of gene mutations identified by whole-exome sequencing

	RAS pathway mutations								Other somatic mutations			
	Somatic				Germline							
Subject number	Gene	Change at DNA level	Change at protein level	VAF[a]	Gene	Change at DNA level	Change at protein level	VAF[a]	Gene	Change at DNA level	Change at protein level	VAF[a]
11[b]	*NF1*	c.4537C>T	p.Arg1513*	40.1/24.2	*NF1*	c.5927delG	p.Trp1976fs	44.0/47.1	*SETBP1*	c.2602G>A	p.Asp868Asn	32.6/27.0
63	*KRAS*	c.38G>A	p.Gly13Asp	44.3/0.0	–	–	–	–	–	–	–	–
72	*PTPN11*	c.172A>T	p.Asn58Tyr	48.2/5.7	–	–	–	–	*SETBP1*	c.2602G>A	p.Asp868Asn	45.9/2.5
									JAK3	c.1970G>A	p.Arg657Gln	30.5/2.2
									SH3BP1	c.830C>T	p.Ser277Leu	47.8/5.1
77	–	–	–	–	–	–	–	–	*SETBP1*	c.2602G>A	p.Asp868Asn	33.4/2.1
78	*NRAS*	c.35G>C	p.Gly12Ala	45.5/9.5	–	–	–	–	–	–	–	–
82	–	–	–	–	*CBL*	c.1217del22	p.Thr406fs	34.7/38.9	–	–	–	–
83	–	–	–	–	*NF1*	c.4970A>G	p.Tyr1657Cys	50.0/51.0	–	–	–	–
84	–	–	–	–	*CBL*	c.1096–110del643	p.Glu366_Phe488del	NA/NA	–	–	–	–
85	*PTPN11*	c.226G>A	p.Glu76Lys	47.5/4.4	–	–	–	–	–	–	–	–
86	*KRAS*	c.38G>A	p.Gly13Asp	38.9/3.1	–	–	–	–	–	–	–	–
89[c]	–	–	–	–	*PTPN11*	c.1502T>G	p.Ser502Ala	50.0/49.9	–	–	–	–
91[c]	–	–	–	–	*PTPN11*	c.218C>T	p.Thr73Ile	49.0/48.0	–	–	–	–
92[c]	–	–	–	–	–	–	–	–	–	–	–	–

NA, not available.
[a]Variant allele frequency (VAF) in tumor/reference samples, where the reference was CD3+ T cells, except for subject 63, for whom umbilical cord was used as the reference. [b]Substantial contamination of tumor cell components in the CD3+ T cell reference. [c]Noonan syndrome–associated myeloproliferative disorder.

translocations that result in an aberrant fusion gene (*NUP98-SETBP1*) and overexpression of *SETBP1* in T cell acute lymphoblastic leukemia (T-ALL) and acute myeloid leukemia (AML), respectively[12,13]. *SETBP1* is one of the downstream targets induced by the Evi-1 oncoprotein[14] and, together with *EVI1* and its homolog *PRDM16* (also known as *MEL1*), was reported to be activated through retrovirus integration. SETBP1 is also known to augment the recovery of granulopoiesis after gene therapies for chronic granulomatous disease[15]. *SETBP1* overexpression is found in more than 27% of adult AML cases and is associated with poor survival[13]. The discovery of recurrent hotspot mutations of *SETBP1* provides unequivocal evidence for the leukemogenic role of deregulated SETBP1 function. Notably, the *SETBP1* mutation encoding p.Asp868Asn was identical to one of the *de novo* mutations reported to be causative in Schinzel-Giedion syndrome (SGS; MIM 269150), which is a highly recognizable congenital disease characterized by severe mental retardation, distinctive facial features and

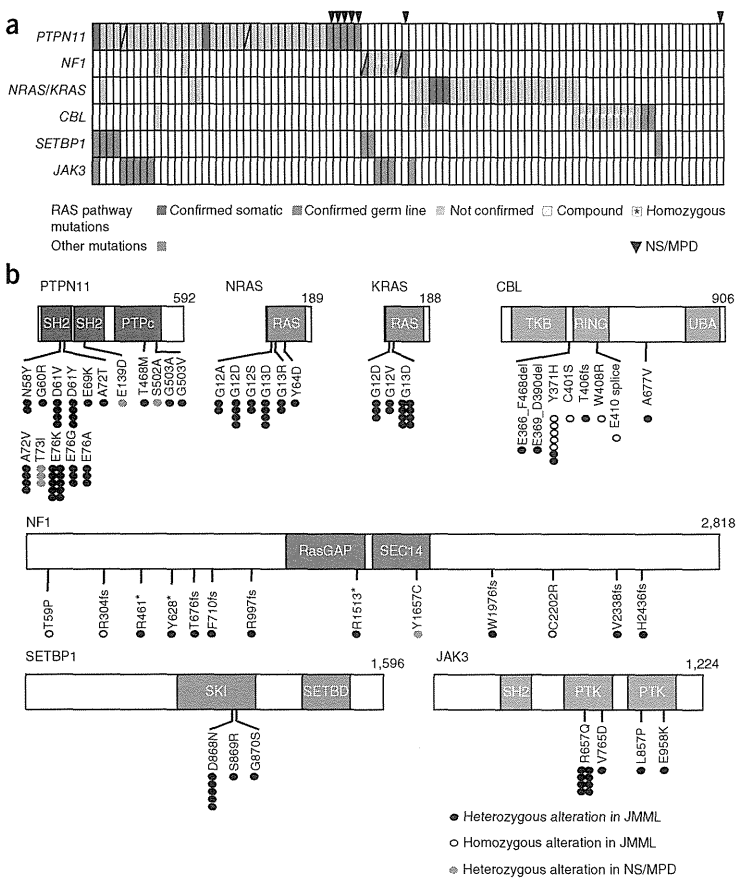

Figure 1 Mutation profiles of 92 JMML cases. (**a**) The mutation status of RAS pathway genes and 2 newly identified gene targets in a cohort of 92 JMML cases is summarized. NS/MPD, Noonan syndrome–associated myeloproliferative disorder. (**b**) The distribution of alterations is shown for each protein. SH2, Src homology 2 domain; PTPc, protein tyrosine phosphatase, catalytic domain; RAS, Ras GTPase family domain; TKB, tyrosine kinase–binding domain; RING, RING-finger domain; UBA, ubiquitin-associated domain; RasGAP, a region of similarity with the catalytic domain of the mammalian p120RasGAP protein in neurofibromin; SEC14, Sec14p-like lipid-binding domain; SKI, v-ski sarcoma viral oncogene homolog domain; SETBD, SET-binding domain; PTK, pseudokinase domain of the protein tyrosine kinases.

Table 2 Subject characteristics

Characteristic	Total cohort (n = 92)	Secondary mutations		P value
		Yes (n = 16)	No (n = 76)	
Sex (male/female)	61/31	12/4	49/27	NS
Median age at diagnosis in months (range)	19 (1–160)	38 (2–160)	13 (1–79)	<0.001
Diagnosis				
JMML	85	16	69	
NS/MPD	7	0	7	
Genetic mutations in RAS pathway				
PTPN11	39	9	30	NS
NF1	9	5	4	0.001
RAS (*NRAS* or *KRAS*)	28 (15/13)	2 (1/1)	26 (14/12)	0.08
CBL	14	0	14	0.06
Without RAS pathway mutation	10	1	9	NS
Secondary genetic mutations				
SETBP1	7	7	0	
JAK3	10	10	0	
Cytogenetics				
Normal karyotype	77	12	65	NS
Monosomy 7	8	1	7	NS
Trisomy 8	4	2	2	NS
Other abnormalities	3	1	2	NS
WBC count at diagnosis $\times 10^9$/l, median (range)	30.0 (1.0–563)	29.6 (5.6–563)	30.0 (1.0–131)	NS
Monocyte count at diagnosis $\times 10^9$/l, median (range)	4.6 (0.2–31.6)	3.1 (0.5–15.2)	4.9 (0.2–31.6)	NS
Percent HbF at diagnosis, median (range)	21 (0–68)	26 (9–55)	16 (0–68)	NS
PLT at diagnosis $\times 10^9$/l, median (range)	61.0 (1.4–483)	47.5 (1.4–175)	65.0 (5.0–483)	NS
HSCT (+/–)	56/36	16/0	40/36	
Alive/deceased	62/30	7/9	55/21	
Percent probability of 5-year overall survival (95% CI)	60 (46–71)	33 (10–59)	65 (49–77)	0.10
Percent probability of 5-year transplantation-free survival (95% CI)	15 (6–27)	0 (0–0)	18 (8–33)	0.007

JMML, juvenile myelomonocytic leukemia; NS/MPD, Noonan syndrome–associated myeloproliferative disorder; WBC, white blood cell; HbF, hemoglobin F; HSCT, hematopoietic stem cell transplantation; NS, not significant. We compared the difference between the subjects with and without secondary mutation, and P values were calculated by two-sided Fisher's exact test or Mann-Whitney U test.

multiple congenital malformations. Individuals with SGS with this mutation have a higher than normal prevalence of tumors, including of neuroepithelial neoplasia[16], although development of myeloid malignancies has not been reported so far.

To further validate our findings, we screened the entire cohort of 92 JMML cases for gene mutations in the newly identified 3 genes together with known RAS pathway targets using deep sequencing[17] (**Supplementary Fig. 5**).

RAS pathway mutations were found in 82 of 92 cases (89%) in a mutually exclusive manner, with *PTPN11* mutations predominant, followed by *NRAS*, *KRAS*, *CBL* and *NF1* mutations (**Fig. 1a** and **Table 2**). In accordance with previous reports, most of the *CBL* (8/14) and *NF1* (4/9) mutations were biallelic (**Fig. 1a,b** and **Supplementary Table 2**)[2,3,18], whereas the majority of mutations in *PTPN11*, *NRAS* and *KRAS* were heterozygous[4]. The individuals without RAS pathway mutations (n = 10) were vigorously investigated by whole-genome sequencing of tumor-normal paired samples (n = 2; **Supplementary Fig. 6**) or by whole-exome sequencing of only tumor samples (n = 8; **Supplementary Fig. 7**). As anticipated, we found no known RAS pathway mutations.

On the other hand, 18 mutations were found in *SETBP1* (n = 7) or *JAK3* (n = 11) in 16 cases (**Fig. 1a,b**, **Table 2** and **Supplementary Table 2**), with these mutations more frequent in cases with mutated *PTPN11* (and possibly *NF1*) than in cases with mutated *NRAS*, *KRAS*

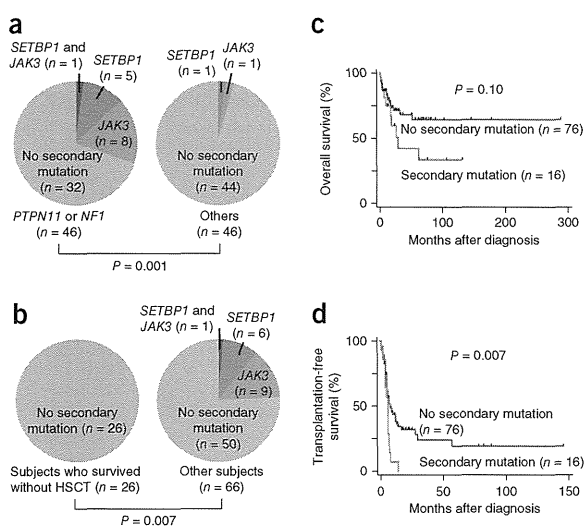

Figure 2 Clinical features of JMML cases with or without secondary mutations. (**a**,**b**) Frequency of secondary mutations in individuals with JMML depending on the type of RAS pathway mutations (left, *PTPN11* or *NF1*; right, other or no mutations) (**a**) and the status of HSCT (**b**). P values were calculated by two-sided Fisher's exact test. (**c**,**d**) The impact of secondary mutations on overall (**c**) and transplantation-free (**d**) survival is shown in Kaplan-Meier survival curves, where statistical significance was tested by log-rank test.

or *CBL* (**Fig. 2a**). Mutations in *SH3BP1*, encoding SH3 domain-binding protein 1, were not recurrent. All *SETBP1* mutations were heterozygous and occurred within the portion of the gene encoding the SKI domain, with six identical to the *de novo* recurrent mutations reported in SGS and five identical to the mutation encoding the p.Asp868Asn alteration (**Fig. 1b**). RT-PCR analysis showed that the wild-type and mutant alleles of *SETBP1* were equally expressed (**Supplementary Fig. 8**). Similarly, 8 of the 11 *JAK3* mutations in 10 cases were the well-described activating mutation (encoding a p.Arg657Gln alteration) found in various hematological malignancies, including Down syndrome–associated acute megakaryoblastic leukemia[19–23], ALL[24,25] and natural killer (NK)/T cell lymphoma[26], and the remaining 3 were also within the portions of the gene encoding the pseudokinase or kinase domain, suggestive of gain of function.

Deep sequencing of the relevant mutant alleles enabled an accurate estimation of allele frequencies for individual mutations (**Supplementary Fig. 9**). *SETBP1* and *JAK3* mutations showed lower allele frequencies (but not with statistical significance for *SETBP1*) than did the corresponding RAS pathway mutations (**Supplementary Fig. 10a**), indicating that the former mutations represent secondary genetic hits that contributed to clonal evolution after the main tumor population was established (**Supplementary Fig. 10b**). Individuals with secondary mutations had shorter lengths of survival compared to those without mutations: 5-year overall survival (hazards ratio (HR) = 1.90, 95% CI = 0.87–4.19). In addition, none of the individuals with JMML who survived without hematopoietic stem cell transplantation (HSCT; $n = 26$) harbored any of the secondary mutations, and individuals with secondary mutations showed significantly inferior 5-year transplant-free survival (HR = 2.18, 95% CI = 1.18–4.02) (**Fig. 2b–d** and **Table 2**).

JMML is characterized by a paucity of gene mutations. The average number of mutations per sample (0.85; range of 0–4) was unexpectedly low compared to those reported in other human cancers (**Supplementary Fig. 11**); excluding common RAS pathway mutations, only 5 mutations were detected in 3 of the 13 discovery cases. This small number of mutations is only comparable to the figure reported for retinoblastoma (mean of 3.3 per case; range of 0–5) (ref. 27) and is in stark contrast to the abundance of gene mutations in chronic myelomonocytic leukemia (CMML) in adult cases, where the mean number of non-silent mutations was 12.4 per sample, of which 3.1 represented known driver changes (ref. 17 and K.Y., M.S., Y.S., D. Nowak, Y. Nagata *et al.*, unpublished data), underscoring the distinct pathogenesis in these two neoplasms that show indistinguishable morphology. The impact of germline events is underscored by the fact that 6 of the 13 discovery cases harbored germline RAS pathway mutations and an additional case without known RAS pathway mutations showed constitutive abnormalities similar to Noonan syndrome. Despite the central role of RAS pathway mutations, a small subset of cases had no documented RAS pathway mutations, even after whole-exome analysis in the two RAS pathway mutation–negative cases, raising the possibility that the latter cases represent a genetically distinct myeloproliferative neoplasm in childhood.

Another key finding in the current study is the discovery of secondary mutations that involve *SETBP1* and *JAK3*. Detected only in a subpopulation of leukemic cells, most of these mutations are thought to be involved in the progression rather than the establishment of JMML and were associated with poor clinical outcome. *SETBP1* is a newly identified proto-oncogene, and identical mutations in this gene have recently been reported in 15–25% of adult cases with atypical chronic myeloid leukemia (CML)[10], CMML and secondary AML[28]. Affecting one of three highly conserved amino acid positions, *SETBP1* mutations have been shown to abolish the binding of an E3 ubiquitin ligase (β-TrCP1) to SETBP1, which prevents ubiquitination and subsequent degradation, leading to gain of function through the consequent increase in SETBP1 protein amounts[10,28]. Although the precise leukemogenic mechanisms of *SETBP1* mutations are still unclear, we have shown that mutant *SETBP1* alleles confer self-renewal capability to myeloid progenitors *in vitro*, and *SETBP1* mutations in adult leukemia were associated with increases in *HOXA9* and *HOXA10* expression[28]. Recurrent *JAK3* mutations in JMML are also noteworthy. The JAK-STAT pathway is a key component of normal hematopoiesis[29]. As in other hematopoietic malignancies[20], the p.Arg657Gln alteration represents the most frequent change in JMML. This alteration confers interleukin (IL)-3 independence to Ba/F3 cells and induces STAT5 phosphorylation[20]. Targeting the JAK-STAT pathway with a pan-JAK inhibitor such as CP-690550 (ref. 30) could be a promising therapeutic possibility for patients with *JAK3*-mutated JMML.

In conclusion, our whole-exome sequencing analysis identified the spectrum of gene mutations in JMML. Together with the high frequency of RAS pathway mutations, the paucity of non–RAS pathway mutations is a prominent feature of JMML. Mutations of *SETBP1* and *JAK3* were common recurrent secondary events presumed to be involved in tumor progression and were associated with poor clinical outcomes. Our findings provide an important clue to understanding the pathogenesis of JMML that may help in the development of novel diagnostics and therapeutics for this leukemia.

URLs. Genomon, http://genomon.hgc.jp/exome/en/; BioCarta, http://www.biocarta.com/; dbSNP131, http://www.ncbi.nlm.nih.gov/projects/SNP/; RefSeq database, http://www.ncbi.nlm.nih.gov/RefSeq/.

METHODS

Methods and any associated references are available in the online version of the paper.

Accession code. We deposited whole-genome and whole-exome sequence data in the European Genome-phenome Archive under accession EGAS00001000521.

Note: Supplementary information is available in the online version of the paper.

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AUTHOR CONTRIBUTIONS

H.S., Y.O., H. Muramatsu, K.Y., M.T., A.K. and M.S. designed and performed the research, analyzed the data and wrote the manuscript. Y.S., K.C., H.T. and S.M. performed bioinformatics analyses of the resequencing data. X.W. and Y.X. performed Sanger sequencing. S.D., A.H., K.N., Y.T. and N.Y. collected specimens and performed the research. H. Makishima and J.P.M. designed the research and analyzed the data. S.O. and S.K. led the entire project and wrote the manuscript.

COMPETING FINANCIAL INTERESTS

The authors declare no competing financial interests.

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