

ORIGINAL ARTICLE

Detection of Allergy-Related *TPSAB1* Copy Number Variation by MALDI-TOF Nucleic Acid Mass Spectrometry

Yang Li ^{1,*}, Xuefeng Zhao ^{2,*}, Sheng He ³, Yunli Lai ¹, Shijie Wei ¹, Wanjun Zhou ¹

* Yang Li and Xuefeng Zhao contributed equally to this work

¹ Department of Medical Genetics, School of Basic Medical Sciences, Southern Medical University, Guangzhou, China

² Department of Laboratory Medicine, Guangming Maternal and Child Health Hospital, Shengzhen, China

³ Guangxi Clinical Research Center for Birth Defects, Guangxi Key Laboratory of Birth Defects Research and Prevention, Guangxi Key Laboratory of Reproductive Health and Birth Defects Prevention, Maternal and Child Health Hospital of Guangxi Zhuang Autonomous Region, Nanning, China

ABSTRACT

Background: Allergic diseases have been linked to genetic variations in *TPSAB1* and *TPSB2*. This study aimed to develop a Matrix-Assisted Laser Desorption/Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS) method for simultaneous copy number detection of *TPSAB1* and *TPSB2*.

Methods: We established a multiplex PCR-tandem MALDI-TOF MS assay for *TPSAB1* and *TPSB2* copy number analysis. To validate the method, 35 standard samples pre-validated by droplet digital PCR (ddPCR) were used to assess accuracy, sensitivity, repeatability, and stability. Additionally, we applied the method to 274 randomly selected population samples for clinical evaluation.

Results: The developed MALDI-TOF MS assay demonstrated high reliability, achieving 100% accuracy in standard samples. The detection limit for the standard template was 20 ng, with repeatability (CV: 8.41 - 13.60%) and stability (CV: 7.75 - 17.38%) within acceptable ranges. In the population cohort, the carrier rate for *TPSAB1* copy number gain (> 2 copies) was 12.41%.

Conclusions: The MALDI-TOF MS-based method is a robust, cost-effective, and high-throughput molecular diagnostic tool, particularly suited for differentiating highly homologous TPS genes. Its high accuracy and reliability make it a promising clinical assay for *TPSAB1* and *TPSB2* copy number analysis.

(Clin. Lab. 2026;72:xx-xx. DOI: 10.7754/Clin.Lab.2025.250701)

Correspondence:

Wanjun Zhou
Department of Medical Genetics
School of Basic Medical Sciences
Southern Medical University
Guangzhou
China
Email: zhouwanjun72@163.com

Supplementary Data

Table S1. Demographic data.

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
1	male	3 years 4 months	healthy	2/0
2	female	5 years 9 months	healthy	2/2
3	female	3 years 10 months	healthy	2/2
4	male	3 years 10 months	healthy	2/2
5	male	5 years 4 months	healthy	2/2
6	male	3 years 8 months	healthy	2/2
7	female	3 years	healthy	2/2
8	female	5 years 10 months	healthy	2/2
9	female	3 years 5 months	healthy	2/2
10	female	3 years 4 months	healthy	4/2
11	male	5 years 4 months	healthy	3/2
12	male	5 years 3 months	healthy	2/2
13	female	3 years	healthy	2/3
14	female	4 years 7 months	healthy	2/2
15	female	4 years 10 months	healthy	2/2
16	male	4 years	healthy	4/2
17	female	4 years 2 months	healthy	4/2
18	female	4 years 9 months	healthy	2/2
19	female	3 years 10 months	healthy	4/2
20	female	3 years 2 months	healthy	2/2
21	male	3 years 3 months	healthy	2/2
22	female	3 years 4 months	healthy	2/2
23	male	4 years 10 months	healthy	2/2
24	female	3 years	healthy	2/2
25	female	3 years 10 months	healthy	2/2
26	female	4 years 5 months	healthy	2/2
27	male	3 years 2 months	healthy	2/2
28	male	3 years	healthy	2/2
29	male	3 years 10 months	healthy	3/2
30	male	3 years 8 months	healthy	2/2
31	male	4 years	healthy	2/2
32	male	4 years 2 months	healthy	2/2
33	male	4 years 2 months	healthy	2/2
34	male	4 years 4 months	healthy	2/2
35	male	3 years 7 months	healthy	2/2
36	male	3 years 2 months	healthy	2/2
37	male	5 years 3 months	healthy	2/2
38	male	3 years 1 months	healthy	2/2
39	male	5 years	healthy	2/2
40	female	4 years 8 months	healthy	2/2
41	female	3 years 7 months	healthy	2/2
42	male	3 years 2 months	healthy	2/2
43	female	2 years 10 months	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
44	male	3 years 7 months	healthy	2/2
45	male	3 years 5 months	healthy	3/2
46	male	3 years 5 months	healthy	2/2
47	male	4 years	healthy	2/2
48	female	3 years 8 months	healthy	2/2
49	female	3 years 4 months	healthy	2/2
50	female	5 years 7 months	healthy	2/2
51	female	2 years 11 months	healthy	2/1
52	male	5 years 8 months	healthy	3/2
53	female	5 years 8 months	healthy	2/2
54	female	4 years	healthy	2/2
55	male	5 years 1 months	healthy	2/2
56	female	3 years	healthy	2/2
57	female	6 years 5 months	healthy	2/2
58	female	3 years 4 months	healthy	2/2
59	male	5 years 8 months	healthy	2/2
60	female	4 years 10 months	healthy	2/2
61	male	2 years 11 months	healthy	2/2
62	male	3 years 11 months	healthy	2/2
63	female	3 years	healthy	2/2
64	male	3 years 8 months	healthy	2/2
65	male	3 years 8 months	healthy	2/2
66	male	3 years 8 months	healthy	2/2
67	female	5 years 8 months	healthy	2/2
68	male	4 years 6 months	healthy	2/2
69	male	4 years 6 months	healthy	2/2
70	male	3 years 7 months	healthy	2/2
71	male	3 years 3 months	healthy	2/2
72	female	3 years 10 months	healthy	2/2
73	male	3 years 2 months	healthy	3/2
74	male	2 years 11 months	healthy	2/0
75	female	5 years 8 months	healthy	2/2
76	female	3 years 4 months	healthy	3/2
77	male	5 years 10 months	healthy	2/2
78	female	4 years	healthy	2/2
79	female	3 years 5 months	healthy	2/2
80	male	6 years 3 months	healthy	2/2
81	male	5 years 1 months	healthy	2/2
82	male	5 years 6 months	healthy	2/2
83	male	3 years 1 months	healthy	2/2
84	female	5 years 1 month	healthy	2/1
85	male	5 years 10 months	healthy	2/2
86	male	4 years 5 months	healthy	2/2
87	female	3 years 1 month	healthy	2/1
88	male	3 years 3 months	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
89	female	3 years 4 months	healthy	2/2
90	male	3 years 11 months	healthy	2/2
91	female	3 years 4 months	healthy	3/2
92	male	5 years 5 months	healthy	3/2
93	male	4 years 5 months	healthy	4/2
94	male	3 years 11 months	healthy	2/2
95	female	4 years 8 months	healthy	2/0
96	male	4 years 1 month	healthy	2/2
97	female	5 years 10 months	healthy	2/2
98	male	4 years 3 months	healthy	2/2
99	male	3 years 3 months	healthy	2/2
100	male	3 years 1 month	healthy	2/2
101	female	3 years 11 months	healthy	3/2
102	male	3 years 11 months	healthy	3/2
103	female	7 years 10 months	healthy	2/2
104	male	4 years 3 months	healthy	2/2
105	female	6 years 6 months	healthy	2/2
106	male	3 years 4 months	healthy	4/2
107	male	2 years 11 months	healthy	2/2
108	male	5 years 11 months	healthy	2/2
109	male	1 year	healthy	2/2
110	male	3 years 11 months	healthy	3/2
111	male	3 years 8 months	healthy	2/2
112	female	4 years 10 months	healthy	2/2
113	male	4 years	healthy	2/2
114	female	3 years 10 months	healthy	3/2
115	male	4 years 1 month	healthy	2/2
116	male	3 years 7 months	healthy	3/2
117	male	3 years 6 months	healthy	2/2
118	male	4 years 10 months	healthy	2/2
119	male	3 years 3 months	healthy	3/2
120	male	3 years 8 months	healthy	2/2
121	female	5 years 3 months	healthy	2/0
122	male	3 years 9 months	healthy	3/2
123	male	3 years 10 months	healthy	2/2
124	female	2 years 11 months	healthy	2/2
125	female	3 years 9 months	healthy	2/2
126	female	3 years 10 months	healthy	3/2
127	male	3 years 7 months	healthy	2/2
128	female	3 years 11 months	healthy	2/2
129	female	3 years 8 months	healthy	2/2
130	female	4 years 5 months	healthy	2/2
131	female	4 years 2 months	healthy	2/2
132	female	3 years 8 months	healthy	2/2
133	female	4 years 2 months	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
134	female	6 years 11 months	healthy	2/1
135	female	3 years 4 months	healthy	2/2
136	female	3 years 10 months	healthy	2/2
137	female	3 years 8 months	healthy	2/2
138	female	5 years 11 months	healthy	3/2
139	female	5 years 11 months	healthy	2/2
140	female	2 years 10 months	healthy	4/2
141	male	4 years 2 months	healthy	2/2
142	male	5 years 4 months	healthy	2/2
143	female	3 years 2 months	healthy	2/2
144	male	4 years 3 months	healthy	2/2
145	female	5 years 1 month	healthy	2/2
146	male	3 years 4 months	healthy	2/2
147	male	3 years	healthy	2/2
148	female	3 years 8 months	healthy	2/2
149	male	3 years 10 months	healthy	2/1
150	female	4 years 8 months	healthy	2/3
151	female	3 years 1 month	healthy	4/2
152	female	5 years 8 months	healthy	2/1
153	male	4 years 10 months	healthy	2/2
154	female	4 years 2 months	healthy	3/2
155	female	4 years 11 months	healthy	2/2
156	male	3 years	healthy	3/2
157	male	5 years 4 months	healthy	2/2
158	male	3 years 4 months	healthy	2/2
159	male	5 years 5 months	healthy	2/2
160	female	3 years 11 months	healthy	2/2
161	male	3 years 6 months	healthy	3/2
162	male	3 years 9 months	healthy	2/2
163	female	3 years 9 months	healthy	2/2
164	male	5 years 3 months	healthy	2/2
165	female	2 years 3 months	healthy	3/2
166	male	5 years 1 month	healthy	2/2
167	female	2 years 9 months	healthy	3/2
168	male	4 years 6 months	healthy	2/1
169	male	3 years 4 months	healthy	4/2
170	female	4 years	healthy	2/2
171	female	3 years	healthy	3/2
172	male	2 years 10 months	healthy	2/2
173	male	4 years 2 months	healthy	2/2
174	female	4 years 11 months	healthy	2/2
175	female	3 years 8 months	healthy	2/2
176	female	3 years 1 month	healthy	2/0
177	female	4 years 3 months	healthy	2/2
178	male	3 years 3 months	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
179	male	5 years 11 months	healthy	2/2
180	female	5 years 7 months	healthy	2/2
181	female	3 years 7 months	healthy	2/2
182	male	3 years 4 months	healthy	3/2
183	female	5 years 11 months	healthy	2/2
184	male	5 years 3 months	healthy	2/2
185	male	3 years 7 months	healthy	2/2
186	female	3 years 2 months	healthy	2/2
187	male	5 years	healthy	2/3
188	male	3 years 10 months	healthy	2/2
189	female	3 years 9 months	healthy	2/2
190	female	3 years 6 months	healthy	2/2
191	female	3 years 5 months	healthy	2/2
192	male	3 years 8 months	healthy	2/2
193	female	3 years 8 months	healthy	2/2
194	female	3 years 7 months	healthy	2/2
195	female	3 years 5 months	healthy	2/2
196	male	5 years 10 months	healthy	2/2
197	female	5 years 6 months	healthy	2/2
198	male	3 years 4 months	healthy	2/3
199	female	5 years 7 months	healthy	2/2
200	male	4 years 6 months	healthy	4/2
201	male	3 years 1 month	healthy	2/2
202	female	3 years 8 months	healthy	2/2
203	female	3 years 10 months	healthy	2/2
204	male	3 years 11 months	healthy	2/2
205	male	3 years 7 months	healthy	2/2
206	male	3 years 5 months	healthy	2/2
207	female	4 years 5 months	healthy	2/2
208	male	3 years 11 months	healthy	2/2
209	male	1 year	healthy	2/2
210	female	3 years 8 months	healthy	2/2
211	male	4 years 2 months	healthy	2/2
212	male	3 years 4 months	healthy	2/0
213	female	3 years 4 months	healthy	2/2
214	female	3 years 9 months	healthy	2/2
215	male	3 years 11 months	healthy	2/2
216	male	3 years	healthy	2/2
217	female	3 years 8 months	healthy	2/2
218	male	5 years 11 months	healthy	2/2
219	male	3 years 5 months	healthy	2/2
220	male	4 years 11 months	healthy	2/2
221	female	3 years 4 months	healthy	2/2
222	female	4 years 10 months	healthy	2/2
223	female	5 years 8 months	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	TPSAB1/TPSB2
224	male	4 years 11 months	healthy	2/2
225	female	3 years 6 months	healthy	2/2
226	male	5 years 3 months	healthy	2/2
227	female	3 years 2 months	healthy	2/2
228	male	3 years 7 months	healthy	2/2
229	female	5 years 11 months	healthy	2/2
230	female	4 years 2 months	healthy	2/2
231	male	5 years 6 months	healthy	2/2
232	male	3 years	healthy	2/3
233	male	3 years 9 months	healthy	2/2
234	male	5 years 5 months	healthy	2/3
235	female	5 years 9 months	healthy	2/2
236	female	3 years 9 months	healthy	2/0
237	male	3 years 4 months	healthy	2/2
238	male	4 years 9 months	healthy	2/2
239	female	4 years	healthy	2/2
240	male	4 years 9 months	healthy	2/2
241	male	3 years	healthy	2/2
242	female	3 years 5 months	healthy	2/2
243	female	3 years 5 months	healthy	2/1
244	male	5 years 8 months	healthy	2/2
245	male	5 years 7 months	healthy	2/2
246	male	3 years 9 months	healthy	2/3
247	male	4 years	healthy	2/2
248	male	3 years 11 months	healthy	2/3
249	female	3 years 9 months	healthy	2/2
250	male	4 years 9 months	healthy	2/3
251	female	3 years 11 months	healthy	2/2
252	female	4 years 2 months	healthy	2/2
253	male	3 years 11 months	healthy	2/2
254	female	3 years 2 months	healthy	2/2
255	female	6 years 7 months	healthy	2/2
256	male	4 years 7 months	healthy	2/3
257	male	4 years	healthy	2/3
258	female	4 years 11 months	healthy	2/2
259	female	4 years 5 months	healthy	2/2
260	female	3 years	healthy	2/2
261	female	3 years 11 months	healthy	2/2
262	male	3 years 6 months	healthy	2/2
263	male	5 years 4 months	healthy	2/2
264	female	5 years 2 months	healthy	2/2
265	male	3 years 8 months	healthy	2/2
266	male	4 years 3 months	healthy	2/2
267	female	3 years 1 months	healthy	2/2
268	male	4 years	healthy	2/2

Table S1. Demographic data (continued).

Serial number	Gender	Age	Healthy status	<i>TPSAB1/TPSB2</i>
269	female	5 years 9 months	healthy	2/2
270	female	3 years 5 months	healthy	2/2
271	female	3 years 4 months	healthy	2/2
272	female	3 years 7 months	healthy	2/2
273	male	3 years 9 months	healthy	2/2
274	male	3 years 4 months	healthy	2/2