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Supplemental Data

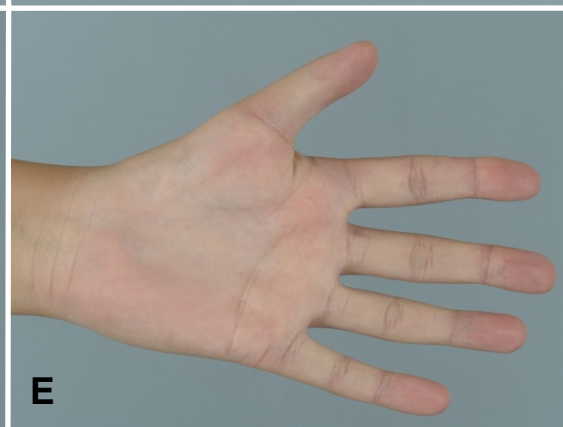
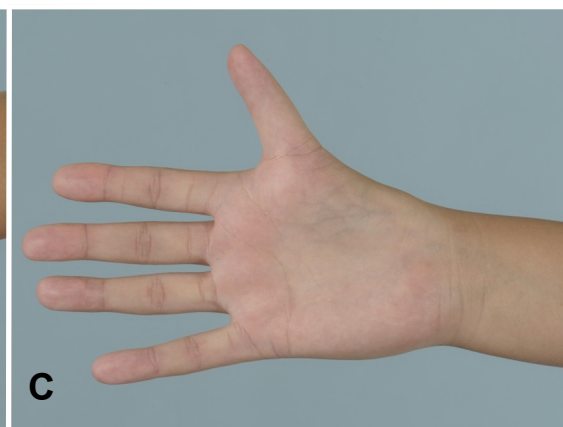
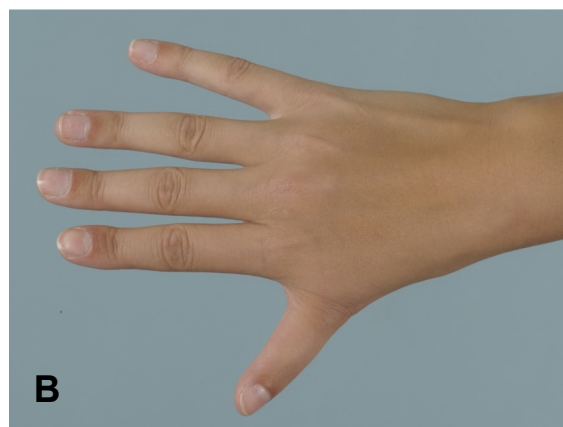
## **Loss-of-Function Mutations in the WNT Co-receptor**

### ***LRP6* Cause Autosomal-Dominant Oligodontia**

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**Figure S1.** Panoramic radiograph of subject F1-III-2 showing taurodontism of the first molars in the upper jaw. The following teeth are agenetic: 18, 15, 12, 22, 28, 38, 35, 33, 43, 45 and 48.



**Figure S2.** Photographs of minor anatomical variation of the ear and underdevelopment of the thumb of subject F3-II-1. A. Ear with localized underdeveloped helix and underdeveloped attached lobe (F3-II-1). B. Hand with underdevelopment of the thumb, proximal placement, right (F3-II-1) C. Hand with underdevelopment thumb and thenar, with decreased palmar creases and absent thenar transverse crease, right (F3-II-1) D. Hand with proximal placement thumb, left (F3-II-1) E. Hand with proximal placement thumb, underdevelopment thenar and shortened proximal transverse crease and absent thenar transverse crease, right (F3-II-1).

				upper right								upper left								
Patient	LRP6 Variant	Gender	Number of missing teeth	18	17	16	15	14	13	12	11	21	22	23	24	25	26	27	28	Taurodontia
F1-II, 1	c.56C>T	m	6	x			x			x			x		x				x	
F1-II, 2	c.56C>T	m	4				x			x			x			x				
<b>F1-III, 1</b>	<b>c.56C&gt;T</b>	<b>m</b>	<b>13</b>	<b>x</b>			<b>x</b>		<b>x</b>	<b>x</b>			<b>x</b>	<b>x</b>		<b>x</b>			<b>x</b>	<b>x</b>
F1-III, 2	c.56C>T	m	7	x			x			x			x						x	x
F2-I, 1	Unaffected	m	N/A																	
F2-I, 2	c.1779dupT	f	6	x					x	x			x	x						
<b>F2-II, 1</b>	<b>c.1779dupT</b>	<b>f</b>	<b>20</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>			<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	
F2-II, 2	Unaffected	m	0																	x
F3-I, 1	c.2224_2225dupTT	m	13	x					x	x			x	x	x	x			x	
F3-II, 1	c.2224_2225dupTT	f	16	x	x		x	x	x	x			x	x					x	
<b>F3-II, 2</b>	<b>c.2224_2225dupTT</b>	<b>f</b>	<b>20</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>			<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>
F4-I, 1	Unaffected	m	0																	
F4-I, 2	c.1144_1145dupAG	f	0																	x
<b>F4-II, 1</b>	<b>c.1144_1145dupAG</b>	<b>m</b>	<b>12</b>	<b>x</b>			<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>		<b>x</b>		<b>x</b>		
<b>F4-II, 1</b>	<b>c.1144_1145dupAG</b>	<b>m</b>	<b>12</b>	<b>x</b>			<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>		<b>x</b>		<b>x</b>		
F4-I, 2	c.1144_1145dupAG	f	0	x																x
F4-I, 1	Unaffected	m	0																	
<b>F3-II, 2</b>	<b>c.2224_2225dupTT</b>	<b>f</b>	<b>20</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>
F3-II, 1	c.2224_2225dupTT	f	16	x	x		x	x		x			x	x		x		x	x	
F3-I, 1	c.2224_2225dupTT	m	13	x			x	x		x	x		x	x		x			x	
F2-II, 2	Unaffected	m	0																	
<b>F2-II, 1</b>	<b>c.1779dupT</b>	<b>f</b>	<b>20</b>	<b>x</b>	<b>x</b>		<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>	<b>x</b>		<b>x</b>	<b>x</b>	
F2-I, 2	c.1779dupT	f	6	x			x									x				x
F2-I, 1	Unaffected	m	N/A																	
F1-III, 2	c.56C>T	m	7	x			x		x					x		x			x	x
<b>F1-III, 1</b>	<b>c.56C&gt;T</b>	<b>m</b>	<b>13</b>	<b>x</b>	<b>x</b>		<b>x</b>				<b>x</b>		<b>x</b>	<b>x</b>		<b>x</b>		<b>x</b>	<b>x</b>	<b>x</b>
F1-II, 2	c.56C>T	m	4																	
F1-II, 1	c.56C>T	m	6	x			x													x
				<b>48</b>	<b>47</b>	<b>46</b>	<b>45</b>	<b>44</b>	<b>43</b>	<b>42</b>	<b>41</b>	<b>31</b>	<b>32</b>	<b>33</b>	<b>34</b>	<b>35</b>	<b>36</b>	<b>37</b>	<b>38</b>	
				lower right								lower left								

**Table S1.** Tooth findings in severe hypodontia index cases (in bold) carrying a *LRP6* variant and information on affected or unaffected family members. The third molars are marked in grey and are not taken into account in the total number of missing teeth.