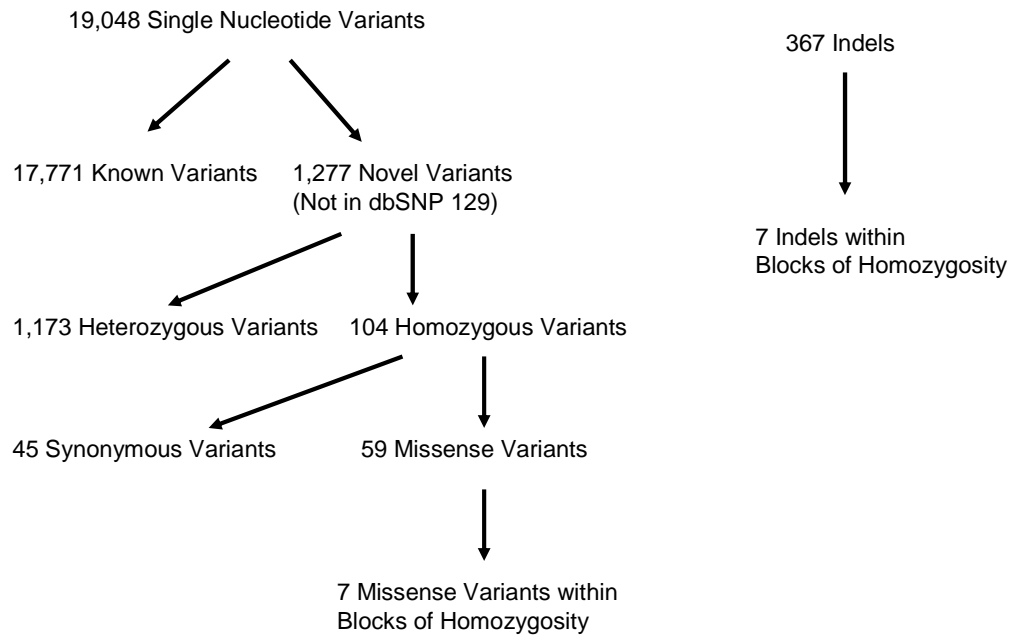
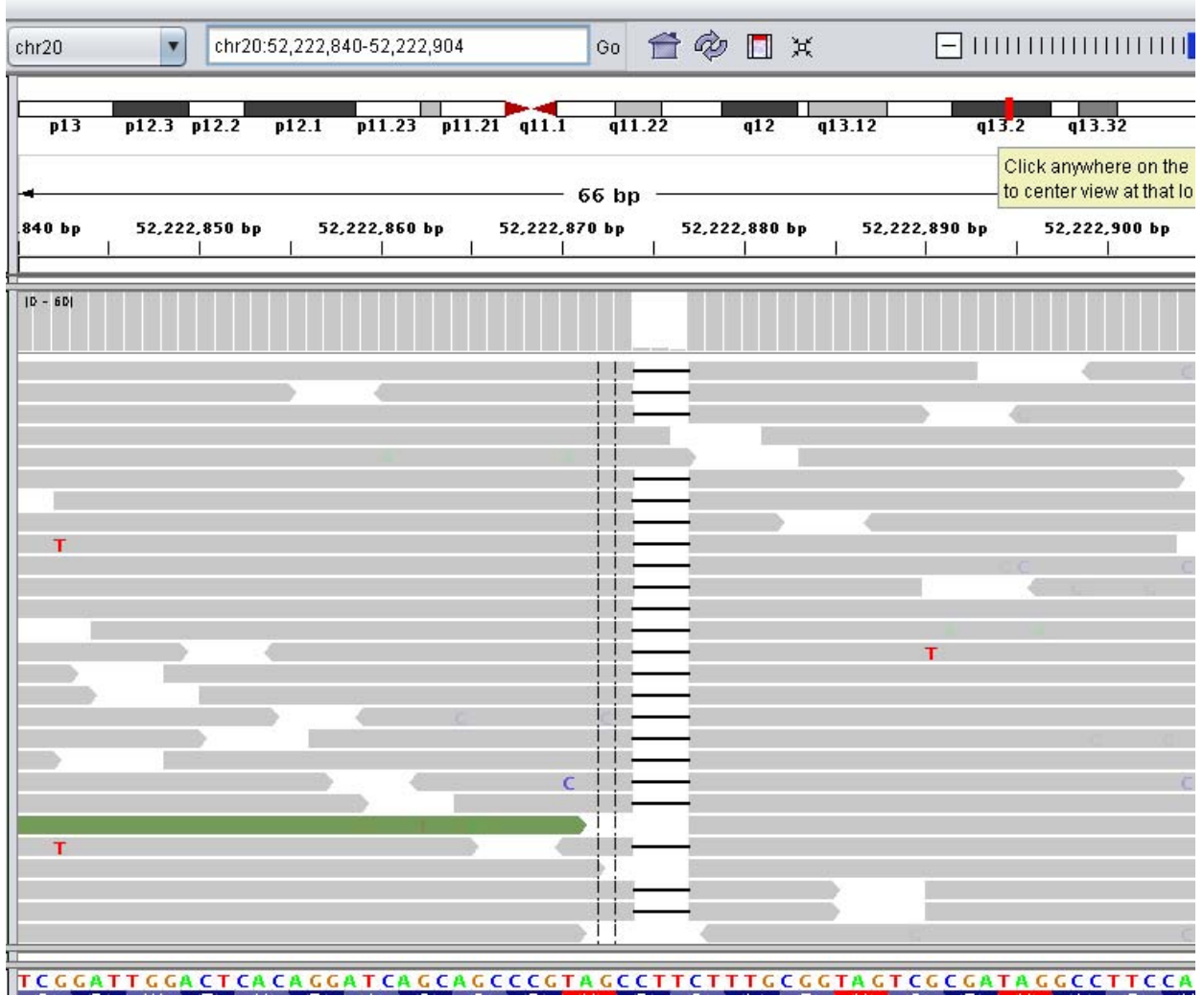


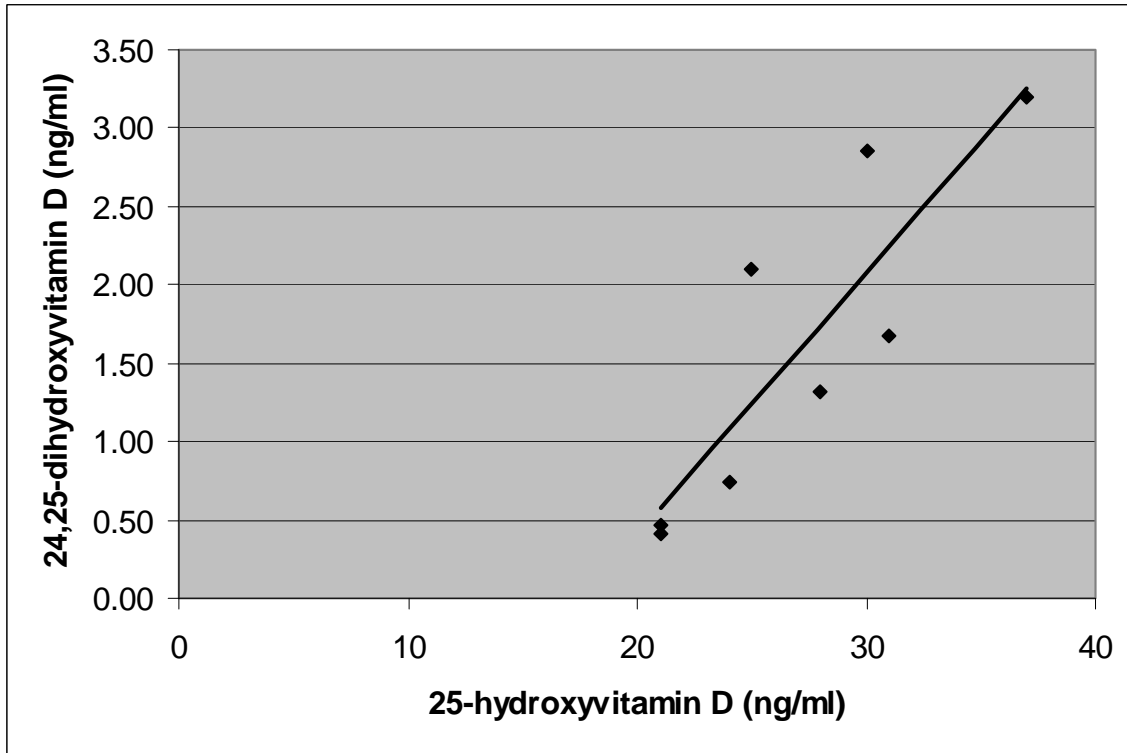
Whole Exome Variant Description



Supplemental Figure 1: Description of variants found in whole exome sequencing of the proband.



Supplemental Figure 2: Visual representation of next generation sequencing data using Integrative Genomics Viewer. The top panel shows the chromosome ideogram with the visualized coordinates below. The grey bars represent individual sequence reads mapped to this region of the genome. The small black lines indicate the 3 base pair deletion found in each read. The reference sequence is present in the bottom panel.



Supplemental Figure 3: Correlation between 24,25-dihydroxyvitamin D and 25-hydroxyvitamin D levels in 8 control subjects ($r=0.86$, $p=0.003$ based on linear regression).

Supplemental Table 1: Individual Biochemical and Radiographic Findings At Presentation of Additional 27 Children (Updated from Reference 6)

Subject ID	Calcium mg/dl (mmol/L)	25(OH)D ng/ml (nmol/L)	1, 25(OH) ₂ D pg/ml (pmol/L)	PTH pg/ml (ng/L)	Urine Ca/Cr mmol/mmol	Nephrocalcinosis
Reference:	8.4 - 10.6 (2.10 - 2.65)	20- 60 (50-150)	15 - 75 (39 - 193)	10 - 65 (10 - 65)	<0.7 (<0.7)	-
5	11.6 (2.90)	36 (91)	8 (22)	1	1.6	+
4	12.6 (3.14)	25 (63)	42 (108)	11	3.6	-*
24	11.3 (2.83)	24 (60)	58 (150)	13	0.5	-
25	11.4 (2.86)	24 (60)	75 (196) ‡	12	1.4	-
32	11.2 (2.79)	18 (46)	77 (200) ‡	1	8.5	+
28	11.2 (2.80)	27 (68)	30 (78)	16	0.3	-
16	10.8 (2.71)	34 (84)	16 (42)	24	0.4	-
27	11.2 (2.79)	33 (83)	20 (53)	36	1.4	+
3	14.1 (3.53)	18 (45)	22 (56)	41	0.7	+
18	11.3 (2.83)	34 (85)	29 (76)	24	1.2	-
13	11.0 (2.75)	22 (54)	32 (83)	31	0.8	-
15	11.6 (2.89)	26 (65)	36 (94)	30	1.5	-
22	11.3 (2.83)	31 (77)	45 (116)	42	1.6	+
26	11.1 (2.78)	17 (43)	45 (117)	34	0.4	N/A
11	10.6 (2.66)	36 (91)	47 (122)	62	0.8	N/A
29	11.9 (2.98)	18 (45)	48 (126)	27	3.3	+
23	11.4 (2.85)	16 (39)	50 (130)	27	1.4	-
20	11.3 (2.82)	13 (33)	51 (132)	50	0.5	+
35	11.0 (2.75)	21 (53)	52 (135)	20	0.6	-
8	10.6 (2.65)	29 (73)	55 (144)	58	0.1	-
31	11.0 (2.76)	N/A	27 (70)	24	1.4	-
7	10.9 (2.73)	28 (70)	62 (160)	28	0.2	N/A
14	10.9 (2.73)	26 (66)	71 (184)	54	0.5	-
10	11.2 (2.79)	51 (127)	75 (196) ‡	33	0.4	+
21	11.3 (2.83)	28 (71)	76 (198) ‡	29	0.3	+
A	19.4 (4.84)	48 (121)	36 (87)	3	N/A	+
B	11.6 (2.89)	65 (162) ‡	53 (127)	9	0.6	+

Legend: ‡ elevated 1, 25(OH)₂D or 25 (OH) Vit D, + nephrocalcinosis, * Renal dysplasia, suppressed PTH, N/A (not available)

Supplemental Table 2: Coordinates of proband's regions of homozygosity. All coordinates are in the in NCBI36/hg18 genome build.

Chromosome	Start Position	End Position	Segment Length
7	139839328	158811981	18972653
16	49075537	64929257	15853720
20	46334251	62382907	16048656