

**Supplemental Table 1:** Biochemical results of CD workup for cases 1 (p.T293I variant) and case 2 (p.G31S variant).

Test results	Case 1	Case 2	Reference range
ACTH (pg/ml)	132	33.6	0-46
Cortisol, 24h urine (mcg/24h)	1835	198	3.5-45
Cortisol levels at 11:30PM and 12AM (mcg/dl)	13.6/13.8	13.3/10.4	<4.4
Cortisol levels at 7:30AM and 8AM (mcg/dl)	28.3/31.6	10.4/9.3	5-25
Serum cortisol (% suppression after 8 mg Dexamethasone (mcg/dl)	1.7 (88%)	2.9 (79%)	(>69% suppression)

CRH test	Case 1		Case 2	
	Cortisol (mcg/dl)	ACTH (pg/ml)	Cortisol (mcg/dl)	ACTH (pg/ml)
minus 5 min	32	147	14.8	43.2
0 min	34.6	139	14.1	34
15 min	37.3	362	23.0	111
30 min	38.9	382	29.4	123
45 min	44.2	343	29.3	171