BETTER LIVING THROUGH GENOMICS

ENDOCRINOLOGY

CLINICAL GENOMICS TEST MENU

ENDOCRINOLOGY

Sl No	TEST NAME	METHOD	SAMPLE TYPE	TAT
1	MODY (Maturity Onset Diabetes of the Young) Panel	NGS	EDTA Blood 3 ml	4 Weeks
2	CAH/ Congenital Adrenal Hyperplasia- 21 hydroxylase deficiency	NGS	EDTA Blood 3 ml	4 Weeks
3	Adrenal Hypoplasia Congenital	NGS	EDTA Blood 3 ml	4 Weeks
4	Combined Pituitary Hormone Deficiency	NGS	EDTA Blood 3 ml	4 Weeks
5	Congenital hypopituitarism gene panel	NGS	EDTA Blood 3 ml	4 Weeks
6	Congenital Hypothyroidism	NGS	EDTA Blood 3 ml	4 Weeks
7	Familial Hypocalcaemia	NGS	EDTA Blood 3 ml	4 Weeks
8	Familial Glucocorticoid Deficiency	NGS	EDTA Blood 3 ml	4 Weeks
9	Hypogonadotropic hypogonadism (including kallamann syndrome)	NGS	EDTA Blood 3 ml	4 Weeks
10	Hypophosphatemia Rickets	NGS	EDTA Blood 3 ml	4 Weeks
11	Hypercholesterolemia gene panel	NGS	EDTA Blood 3 ml	4 Weeks
12	Hyperlipidaemia	NGS	EDTA Blood 3 ml	4 Weeks
13	Multiple Endocrine Neoplasia (MEN1, MEN 2A, 2A, 4)	NGS	EDTA Blood 3 ml	4 Weeks
14	Neonatal Hyperinsulinemia/ Congenital Hyperinsulinemia	NGS	EDTA Blood 3 ml	4 Weeks
15	Obesity (non-syndromic)	NGS	EDTA Blood 3 ml	4 Weeks
16	Obesity (syndromic)	NGS	EDTA Blood 3 ml	4 Weeks
17	Primordial Dwarfism	NGS	EDTA Blood 3 ml	4 Weeks
18	Phaeochromocytoma	NGS	EDTA Blood 3 ml	4 Weeks
19	Pseudo hypoaldosteronism panel	NGS	EDTA Blood 3 ml	4 Weeks
20	Syndromic Short Stature	NGS	EDTA Blood 3 ml	4 Weeks
21	Ciliopathies	NGS	EDTA Blood 3 ml	4 Weeks
22	Glaucoma	NGS	EDTA Blood 3 ml	4 Weeks
23	Cystic Fibrosis	NGS	EDTA Blood 3 ml	4 Weeks
24	Cystic fibrosis (CFTR) del F508 mutation analysis	Sanger	EDTA Blood 3 ml	10 working days