

Endocrinology

SI No.	Test Name	Gene list	Specimen Type	Methodology	TAT
1	MCM6 gene sequencing	MCM6	Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
2	MKRN3 gene sequencing	MKRN3	Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
3	HNF1B gene sequencing	HNF1B	Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
4	Pseudohypoparathyroidism Ia (GNAS) gene sequencing	GNAS	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
5	MC4R gene sequencing	MC4R	Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
6	GATA3 gene sequencing	GATA3	Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
7	NROB1, ABCD1, AIRE gene sequencing	ABCD1,AIRE,NROB1	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
8	CYP11B1 gene sequencing	CYP11B1	DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
9	[Combo] Congenital adrenal hyperplasia CYP21A2 (21-OH) combo - NGS and deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Combo test	14 days
10	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency (HSD3B2) gene analysis		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
11	SRD5A2 gene analysis		Peripheral Blood in EDTA (Purple Top),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
12	Monogenic and syndromic obesity gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
13	Leptin deficiency (LEP) gene analysis		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
14	Alstrom syndrome (ALMS1) gene analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
15	Kallmann syndrome gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
16	Congenital hypopituitarism gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
17	Congenital adrenal hyperplasia CYP21A2 (21-OH) deletion/duplication analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	MLPA	14 days
18	Androgen receptor (AR) gene analysis		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
19	Hereditary pancreatitis gene panel		Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA	Next Generation Sequencing (NGS)	19 days
20	Hypercholesterolemia gene panel		Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top)	Next Generation Sequencing (NGS)	19 days
21	Maturity-Onset Diabetes of the Young (MODY) & neonatal diabetes gene panel		DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF)	Next Generation Sequencing (NGS)	19 days
22	Congenital adrenal hyperplasia CYP21A2 (21-OH) gene analysis		Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS)	Next Generation Sequencing (NGS)	14 days