UO Genetica Medica-Azienda Ospedaliero-Universitaria di Parma (Direttore: Prof. Antonio Percesepe)

Selezionare con una X un pannello se si desidera l'analisi di tutti i geni inclusi, oppure gli specifici geni di interesse.
I geni non selezionati nel pannello richiesto potranno essere analizzati in futuro, in seguito a richiesta specifica.

Per informazioni: Dott. Davide Martorana, Dott.ssa Patrizia Caggiati, Dott.ssa Federica Riccardi, Dott.ssa Beatrice Bortesi (0521-704463)

HEMOPHILIA AND RARE BLEEDING DISORDERS	**F8**	XLR	Hemophilia A
	F9	XLR	Hemophilia B
	F7	AR	Factor VII deficiency
	F11	AD	Factor XI deficiency, autosomal dominant
		AR	Factor XI deficiency, autosomal recessive
	VWF	AD	von Willebrand disease, type 1
		AD/AR	von Willebrand disease, types 2A, 2B, 2M, and 2N
		AR	von Willibrand disease, type 3
	F13A1	AR	Factor XIIIA deficiency
	F13B	AR	Factor XIIIB deficiency

NEUROFIBROMATOSIS – SCHWANNOMATOSIS	**NF1**	AD	Neurofibromatosis type 1
	SPRED1	AD	Legius syndrome
	NF2	AD	Neurofibromatosis type 2
	SMARCB1	AD	Schwannomatosis type 1

HEREDITARY PERIODIC FEVERS – AUTOINFLAMMATORY	**CARD14**	AD	Psoriasis type 2
		AD	Pityriasis rubra pilaris
	AP1S3	AD	Pustular Psoriasis
	C1NH	AR/AD	Angioedema, hereditary, types I and II
		AD	Complement component 4, partial deficiency of
	CECR1 (ADA2)	AR	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
		AR	Sneddon syndrome
	IL1RN	AR	Interleukin 1 receptor antagonist deficiency
	IL36RN	AR	Psoriasis 14, pustular
	LPIN2	AR	Majeed syndrome
	MEFV	AR/AD	Familial Mediterranean Fever
	MVK	AR	Hyper-IgD syndrome
		AR	Mevalonic Aciduria
	NEMO	AD	Osteolysis, familial expansile
	NLRC4	AD	Familial cold autoinflammatory syndrome 4

SYNDROMES	**NLRC4**	AD	Autoinflammation with infantile enterocolitis
	NLRP12	AD	Familial cold autoinflammatory syndrome 2
	NLRP3	AD	CINCA syndrome
		AD	Muckle-Wells syndrome
		AD	Familial cold inflammatory syndrome 1
	NLRP7	AR	Hydatidiform mole, recurrent, 1
	NOD2/CARD15	AD	Blau Syndome
	PLCG2	AD	Autoinflammation, antibody deficiency, and immune dysregulation syndrome
		AD	Familial cold autoinflammatory syndrome 3
	PSMB8	AR	Proteasome-associated autoinflammatory syndrome 1 and digenic forms
	PSTPIP1	AD	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne
	TMEM173	AD	STING-associated vasculopathy, infantile-onset
	TNFRSF11A	AD	Osteolysis, familial expansile
	TNFRSF1A	AD	TNF-Receptor-Associated Periodic Fever Syndrome

CYSTIC FIBROSIS	**CFTR**	AR	Congenital bilateral absence of vas deferens
		AR	Cystic fibrosis
			Sweat chloride elevation without CF

HEREDITARY THORACIC AORTIC ANEURYSM AND DISSECTION	**FBN1**	AD	Acromicric dysplasia
		AD	Aortic aneurysm, ascending, and dissection
		AD	Ectopia lentis, familial
		AD	Geleophysic dysplasia 2
		AD	Marfan lipodystrophy syndrome
		AD	Marfan syndrome
		AD	MASS syndrome
		AD	Stiff skin syndrome
		AD	Weill-Marchesani syndrome 2, dominant
	TGFBR1	AD	Loeys-Dietz syndrome 1
	TGFBR2	AD	Colorectal cancer, hereditary nonpolyposis, type 6
		AD	Esophageal cancer, somatic
		AD	Loeys-Dietz syndrome 2
	SMAD3	AD	Loeys-Dietz syndrome 3
	MYLK	AD	Aortic aneurysm, familial thoracic 7
	ACTA2	AD	Aortic aneurysm, familial thoracic 6
		AD	Moyamoya disease 5
		AD	Multisystemic smooth muscle dysfunction syndrome
	COL3A1	AD	Ehlers-Danlos syndrome, type IV
	TGFB2	AD	Loeys-Dietz syndrome 4
	TGFB3	AD	Arrhythmogenic right ventricular dysplasia 1
		AD	Loeys-Dietz syndrome 5
	MAT2A	AD	thoracic aortic aneurysm
	MFAP5	AD	Aortic aneurysm, familial thoracic 9
	PRKG1	AD	Aortic aneurysm, familial thoracic 8

	AGXT	AR	Hyperoxaluria, primary, type 1

NEFROPATIE A BASE GENETICA	**AQP2**	AD/AR	Diabetes insipidus, nephrogenic, 2
	ATP6V0A4	AR	Distal renal tubular acidosis 3, with or without sensorineural hearing loss
	ATP6V1B1	AR	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss
	AVPR2	XLR	Diabetes insipidus, nephrogenic, 1
		XLR	Nephrogenic syndrome of inappropriate antidiuresis
	BSND	AR	Bartter syndrome, type 4a
		AR	Sensorineural deafness with mild renal dysfunction
	CASR	AD	Hypocalcemia, autosomal dominant, with Bartter syndrome
		AD	Hypocalciuric hypercalcemia, type I
	CEP290	AR	Bardet-Biedl syndrome 14
	CLCN5	XLR	Dent disease 1
		XLR	Nephrolithiasis, type I
		XLR	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis
	CLCNKB	AR	Bartter syndrome, type 3
	COL4A3	AD	Alport syndrome, autosomal dominant
		AR	Alport syndrome, autosomal recessive
		AD	Hematuria, benign familial
	COL4A4	AR	Alport syndrome, autosomal recessive
		AD	Hematuria, familial benign
	COL4A5	XLD	Alport syndrome
	CRB2	AR	Focal segmental glomerulosclerosis 9
		AR	Ventriculomegaly with cystic kidney disease
	CTNS	AR	Cystinosis, atypical nephropathic
		AR	Cystinosis, late-onset juvenile or adolescent nephropathic
		AR	Cystinosis, nephropathic
	CUBN	AR	Proteinuria, chronic benign
	CYP24A1	AR	Hypercalcemia, infantile, 1
	DSTYK	AD	Congenital anomalies of kidney and urinary tract 1
	EMP2	AR	Nephrotic syndrome, type 10
	FN1	AD	Glomerulopathy with fibronectin deposits 2
	GRHPR	AR	Hyperoxaluria, primary, type II
	HNF1b	AD	Renal cysts and diabetes syndrome
	KANK2	AR	Nephrotic syndrome, type 16
	KCNJ1	AR	Bartter syndrome, type 2
	LAMB2		Nephrotic syndrome, type 5, with or without ocular abnormalities
	NPHS2	AR	Nephrotic syndrome, type 2
	OCRL	XLR	Dent disease 2
		XLR	Lowe syndrome
	PAX2	AD	Glomerulosclerosis, focal segmental, 7
		AD	Papillorenal syndrome
	PHEX	XLD	Hypophosphatemic rickets, X-linked dominant
	PKD1	AD	Polycystic kidney disease 1
	PKD2	AD	Polycystic kidney disease 2
	PKHD1	AR	Polycystic kidney disease 4, with or without hepatic disease
	SLC12A1	AR	Bartter syndrome, type 1
	SLC12A3	AR	Gitelman syndrome
	SLC34A1	AD	Nephrolithiasis/osteoporosis, hypophosphatemic, 1
		AR	Hypercalcemia, infantile, 2
	SLC4A1	AD	Distal renal tubular acidosis 1
		AR	Distal renal tubular acidosis 4 with hemolytic anemia

	SLC4A4	AR	Renal tubular acidosis, proximal, with ocular abnormalities
	TTC21B	AD/AR	Nephronophthisis 12
	UMOD	AD	Tubulointerstitial kidney disease, autosomal dominant, 1

HEREDITARY NON-POLYPOSIS COLORECTAL CANCER (HNPCC)	**MLH1**		Colorectal cancer, hereditary nonpolyposis, type 2
	MSH2	AD	Colorectal cancer, hereditary nonpolyposis, type 1
	MSH6	AD	Colorectal cancer, hereditary nonpolyposis, type 5
	PMS2		Colorectal cancer, hereditary nonpolyposis, type 4
	EPCAM	AD	Colorectal cancer, hereditary nonpolyposis, type 8

POLYPOSIS, HAMARTOMATOUS INTESTINAL POLYPS-AND-SPOTS SYNDROME	**STK11**	AD	Peutz-Jeghers syndrome

HEREDITARY DIFFUSE GASTRIC CANCER (HDGC)	**CDH11**	AD	Elsahy-Waters syndrome