A	**ALPORT SYNDROME**	☐	**COL4A3**	AD	Alport syndrome, autosomal dominant
				AR	Alport syndrome, autosomal recessive
				AD	Hematuria, benign familial
		☐	**COL4A4**	AR	Alport syndrome, autosomal recessive
					Hematuria, familial benign
		☐	**COL4A5**	AD	Alport syndrome
B	**HEMOPHILIA AND RARE BLEEDING DISORDERS**	☐	**F8**	XLR	Hemophilia A
		☐	**VWF**	AD	von Willebrand disease, type 1
				AD, AR	von Willebrand disease, types 2A, 2B, 2M, and 2N
				AR	von Willibrand disease, type 3
		☐	**F11**	AD	Factor XI deficiency, autosomal dominant
				AR	Factor XI deficiency, autosomal recessive
		☐	**F7**	AR	Factor VII deficiency
C	**CYSTIC FIBROSIS**	☐	**CFTR**	AR	Congenital bilateral absence of vas deferens
				AR	Cystic fibrosis
					Sweat chloride elevation without CF
D	**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
		☐	**MYH11**	AD	Aortic aneurysm, familial thoracic 4
		☐	**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
		☐	**FLNA**	XLR	Cardiac valvular dysplasia, X-linked
				XLR	Congenital short bowel syndrome
					FG syndrome 2
				XLR	Frontometaphyseal dysplasia 1
				XLD	Heterotopia, periventricular
				XLR	Intestinal pseudoobstruction, neuronal
				XLD	Melnick-Needles syndrome
				XLD	Otopalatodigital syndrome, type I
				XLD	Otopalatodigital syndrome, type II
					Terminal osseous dysplasia
		☐	**NOTCH1**	AD	Adams-Oliver syndrome 5
				AD	Aortic valve disease 1
		☐	**MAT2A**	AD	thoracic aortic aneurysm
		☐	**MFAP5**	AD	Aortic aneurysm, familial thoracic 9
		☐	**PRKG1**	AD	Aortic aneurysm, familial thoracic 8
E	**NEUROFIBROMATOSIS TYPE 1**	☐	**NF1**	AD	Neurofibromatosis type 1
		☐	**SPRED1**	AD	Legius syndrome

Note: 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 potranno essere analizzati in futuro, in seguito a richiesta specifica