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BIOLAB

MATERNITY eKARYO FULL

REPORT V 1.0

GENI ANALIZZATI

Il test Maternity eKaryo Full è un test genetico che consente di conoscere il proprio eventuale stato di portatore sano delle malattie genetiche. Il test è eseguito su un campione di sangue intero o di saliva. Il Test analizza malattie recessive (di cui possiamo essere portatori sani), X-linked (di cui le donne possono essere portatrici sane) e alcune malattie autosomiche dominanti.

A4GALT	ABCD1	ACADM	ACADS	ACADSB	ACADVL	ACAT1	ACE
ADA	AHI1	AIPL1	AK2	ALDH3A2	ALDH5A1	ALDH7A1	ALOX12B
ALOXE3	ARG1	ARHGEF9	ARL13B	ASL	ASS1	ATF6	AUH
B2M	BCKDHA	BCKDHB	BTD	CAT	CBS	CCBE1	CD3D
CD3E	CDH7	CDKL5	CEP41	CERS3	CFTR	CHRNA1	CLDN1
COCH	COL1A1	COL2A1	COL4A3	COL4A4	COL4A5	CPS1	CPT1A
CPT2	CRELD1	CRYM	CTNNA3	CTSA	CYP11A1	CYP11B1	CYP1B1
CYP21A2	CYP24A1	CYP27A1	CYP4F22	DBT	DCLRE1C	DCTN4	DFNA5
DHDDS	DIABLO	DIAPH1	DLD	DNAH5	DNAI1	DNAJC19	DUOX2
EPAS1	EPM2A	ERCC2	ERCC8	ETFA	ETFB	ETFDH	FAH
FAM111A	FGF23	FGFR3	FOXN1	FOXP3	FOXRED1	FRMD7	FZD4
G6PC	G6PD	GAA	GALE	GALK1	GALT	GCDH	GCH1
GIPC3	GJB1	GJB2	GJB3	GJB4	GJB6	GJC2	GLA
GNMT	GRHL2	GUSB	HADH	HADHA	HADHB	HBB	HFE
HLCS	HMGCL	HPD	HSD17B10	HSD3B2	IGHMBP2	IL2RG	IL7R
ITGB3	IVD	JAK3	KCNQ2	KCNQ3	KCNQ4	KRT1	KRT10
KRT2	LAMA2	LIG4	LIPN	LRP5	LTBP2	MAP2K1	MAP2K2
MAT1A	MBTPS2	MCCC1	MCCC2	MEGF10	MID1	MMAA	MMAB
MMACHC	MMADHC	MTHFR	MTR	MTRR	MUT	MYH14	MYO1A
NDP	NDUFA12	NDUFA2	NDUFA9	NDUFAF6	NDUFS7	NDUFS8	NEU1
NF1	NF2	NHEJ1	NHLRC1	NIPAL4	NMNAT1	NRAS	NSDHL
OPA3	OTOA	OTOF	PAH	PAX6	PAX8	PCBD1	PCCA
PCDH19	PCSK5	PDE6A	PDE6B	PHGDH	PIEZO1	PIGL	PKHD1
PLCB1	PLXND1	PNPLA1	POU4F3	PRRT2	PSEN1	PSEN2	PTPN11
PTPRC	PTS	QDPR	RAF1	RAG1	RAG2	RDH12	RECQL4
RFX5	RFXANK	RP1	RP2	RPGR	SCN2A	SCNN1A	SCNN1B
SCNN1G	SCO2	SEMA3E	SFTPB	SFTPC	SHOC2	SIX6	SLC17A8
SLC22A5	SLC25A13	SLC25A20	SLC25A22	SLC27A4	SLC5A5	SNAP29	SOD1
SOD2	SOS1	SPTB	ST14	STIM1	STRA6	STS	STX1A
STXBP5	SYN1	TAT	TAZ	TBCE	TBX20	TCF4	TFRC
TG	TGFB1	TGM1	TH	TLL1	TNNT2	TPO	TRIP11
TSHB	UBE3A	UGT1A1	VLDLR	ZAP70	ZNF469		

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METODI E LIMITI

Il test Maternity eKaryo Full è un test di sequenziamento di ultima generazione (NGS) per il rilevamento di varianti in 254 geni. Il test viene eseguito sulla saliva o sul sangue intero. Il DNA genomico estratto viene processato con analisi ampliseq e sequenziato su sequenziatore ThermoFisher o Illumina. I dati di sequenziamento vengono elaborati utilizzando la pipeline bioinformatica proprietaria e sviluppata per l'uso previsto. La mappatura e l'analisi si basano sulla sequenza di riferimento UCSC hg19 del genoma umano. Le varianti a singolo nucleotide, piccole inserzioni/ delezioni (nei geni selezionati) sono rilevate. Le regioni promotrici sono escluse.Vengono rilevate solo le varianti ereditarie (germinali). Inversioni e complessi riarrangiamenti strutturali come traslocazioni non vengono rilevati. Sono escluse dal report le posizioni con copertura inferiore a 30X a meno che non sia confermata da una tecnologia alternativa. Le varianti vengono classificate come patogeniche, probabilmente patogeniche, di significato incerto (VUS), probabilmente benigne, o benigne, sulla base delle linee guida dell' American College of Medical Genetics and Genomics (ACMG) (PMID: 25741868). Le varianti patogeniche, probabilmente patogeniche, e le VUS sono incluse nel report, mentre le varianti benigne e probabilmente benigne non vengono riportate. Non vengono riportate le varianti introniche. Le varianti patogeniche, probabilmente patogeniche e le VUS rilevate mediante NGS sono confermate attraverso sequenziamento Sanger quando necessario. Test basati su tecnologia NGS potrebbero non essere in grado di rilevare alcune varianti e potrebbero esserci altri geni associati a sindromi tumorali ereditarie che non sono coperte da questo pannello.

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PATOLOGIE ANALIZZATE

Hypothyroidism, congenital, nongoitrous 4	Cardiomyopathy, dilated, 1U	Alzheimer disease, type 3
Alzheimer disease, type 3, with spastic paraparesis and apraxia	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques	Dementia, frontotemporal
Pick disease	Immunodeficiency	Trichothiodystrophy 1, photosensitive;
Xeroderma pigmentosum, group D	Aniridia	Anterior segment dysgenesis 5, multiple subtypes
Cataract with late-onset corneal dystrophy	Foveal hypoplasia 1	Keratitis
Optic nerve hypoplasia	Achondrogenesis, type II or hypochondrogenesis	Avascular necrosis of the femoral head
Czech dysplasia	Kniest dysplasia	Legg-Calve-Perthes disease
Osteoarthritis with mild chondrodysplasia	Platyspondylic skeletal dysplasia, Torrance type	SED congenita
SMED Strudwick type	Spondyloepiphyseal dysplasia	Stickler sydrome, type I
Bronchiectasis with or without elevated sweat chloride 2	Pseudohypoaldosteronism, type I	Ichthyosis follicularis - alopecia - photophobia
Osteogenesis imperfecta, type XIX	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	(Brain and CNS)(Williams syndrome)
Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy.https://pubmed.ncbi.nlm.nih.gov/31494578/	2-methylbutyrylglycinuria	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
3-hydroxyacyl-CoA dehydrogenase deficiency; Hyperinsulinemic hypoglycemia, familial, 4	Hyperinsulinemic hypoglycemia, familial, 4	3-Methylcrotonyl-CoA carboxylase 1 deficiency
3-Methylcrotonyl-CoA carboxylase 2 deficiency	3-methylglutaconic aciduria, type I	3-methylglutaconic aciduria, type III

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Optic atrophy 3 with cataract	3-methylglutaconic aciduria, type V	4-hydroxybutyric aciduria
Acatalasemia	Achondrogenesis, type IA	Odontochondrodysplasia 1
Achondroplasia	CATSHL syndrome	Crouzon syndrome with acanthosis nigricans
Hypochondroplasia	LADD syndrome	Muenke syndrome
severe achondroplasia with developmental delay and acanthosis nigricans SADDAN	Thanatophoric dysplasia, type I	Thanatophoric dysplasia, type II
Achromatopsia 7	Acyl-CoA dehydrogenase, medium chain, deficiency of	"Acyl-CoA dehydrogenase, short-chain, deficiency of "
Adenosine deaminase deficiency, partial	Severe combined immunodeficiency	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
Aldosteronism, glucocorticoid-remediable	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency
Adrenal insufficiency, due to CYP11A1 deficiency	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	Adrenoleukodystrophy
Adrenomyeloneuropathy, adult	Alpha-methylacetoacetic aciduria	Alport syndrome
Hematuria, benign familial	Hematuria, benign familial	Alport syndrome
Alport syndrome X-Linked	Cardiomyopathy, dilated, 1V	Amyotrophic lateral sclerosis 1
Spastic tetraplegia and axial hypotonia, progressive	Elliptocytosis-3	Spherocytosis, type 2
Angelman syndrome	Anterior segment dysgenesis 6, multiple subtypes	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset
Argininemia	Argininosuccinic aciduria	Arrhythmogenic right ventricular dysplasia, familial, 13

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Atrial septal defect 4	Atrial septal defect 6	Atrioventricular septal defect, partial, with heterotaxy syndrome
Autoimmune disease, multisystem, infantile-onset, 2	Immunodeficiency 48	Dopa-responsive dystonia
Baller-Gerold syndrome	RAPADILINO syndrome	Rothmund-Thomson syndrome, type 2
Bare lymphocyte syndrome, type II, complementation group C and group E	Bart-Pumphrey syndrome	Deafness
Hystrix-like ichthyosis with deafness	Keratitis-ichthyosis-deafness syndrome	Keratoderma, palmoplantar, with deafness
Vohwinkel syndrome	Barth syndrome	Biotinidase deficiency
Bleeding disorder, platelet-type, 24,	Glanzmann thrombasthenia 2	Thrombocytopenia, neonatal alloimmune
Brittle cornea syndrome	Bronchiectasis with or without elevated sweat chloride 1	Liddle syndrome 1
Pseudohypoaldosteronism, type I	Bronchiectasis with or without elevated sweat chloride 3	Liddle syndrome 2
Pseudohypoaldosteronism, type I	Caffey disease	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1
Ehlers-Danlos syndrome, arthrochalasia type, 1	Osteogenesis imperfecta, type I, type II, type III, type IV	Camurati-Engelmann disease
Inflammatory bowel disease, immunodeficiency, and encephalopathy	Carbamoylphosphate synthetase I deficiency	"Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase
deficiency 1"	Cardiofaciocutaneous syndrome 3	Cardiofaciocutaneous syndrome 4
Cardiomyopathy, dilated, 1D	Cardiomyopathy, familial restrictive, 3	Cardiomyopathy, hypertrophic, 2
Left ventricular noncompaction 6	Cardiomyopathy, dilated, 1NN	LEOPARD syndrome 2

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Noonan syndrome 5	Carnitine deficiency, systemic primary	Carnitine-acylcarnitine translocase deficiency
Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome	"Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma	syndrome"
Cerebrotendinous xanthomatosis	Charcot-Marie-Tooth disease, axonal, type 2S	Neuronopathy, distal hereditary motor, type VI
Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	CHARGE syndrome	Hypogonadotropic hypogonadism 5 with or without anosmia
CHARGE syndrome;	CHILD syndrome	CK syndrome
CHIME syndrome	Ciliary dyskinesia, primary, 1	Ciliary dyskinesia, primary, 3
Citrullinemia	Citrullinemia, type II, neonatal-onset	Citrullinemia, adult-onset type II
Cockayne syndrome type A	UV-sensitive syndrome 2	Combined cellular and humoral immune defects with granulomas
Omenn syndrome (AR)	Severe combined immunodeficiency, B cell-negative	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity
Combined cellular and humoral immune defects with granulomas	Omenn syndrome	Severe combined immunodeficiency, B cell-negative
Combined immunodeficiency, moderate	Severe combined immunodeficiency,	Cystic fibrosis
Congenital muscular dystrophy type 1A	Convulsions, familial infantile, with paroxysmal choreoathetosis	Episodic kinesigenic dyskinesia 1
Seizures, benign familial infantile, 2	CPT deficiency, hepatic, type IA	CPT II deficiency, infantile
CPT II deficiency, myopathic, stress-induced	Crigler-Najjar syndrome, type I	Crigler-Najjar syndrome, type II

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Hyperbilirubinemia, familial transient neonatal	Deafness	Erythrokeratodermia variabilis et progressiva 1
Deafness	Ectodermal dysplasia 2, Clouston type	Deafness type 1
Deafness type 15	Deafness type 25	Deafness type 28
Deafness type 2A	Deafness type 4	Deafness type 40
Deafness type 48	Deafness type 5	Deafness type 50
Deafness type 64	Deafness type 9	Deafness, autosomal recessive 15
Deafness, autosomal recessive 22	Deafness, autosomal recessive 9	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema
Lymphatic malformation 6	Sickle cell anemia	Thalassemia, beta
Developmental and epileptic encephalopathy 11	Episodic ataxia, type 9	Seizures, benign familial infantile, 3
Developmental and epileptic encephalopathy 7	Myokymia	Seizures, benign neonatal, 1
Dihydrolipoamide dehydrogenase deficiency	docking and/or fusion of synaptic vesicles with the presynaptic plasma membrane	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia
Hyperphenylalaninemia, BH4-deficient, B	Encephalopathy, progressive, with amyotrophy and optic atrophy	Hypoparathyroidism-intellectual deficit-dysmorphism syndrome
Kenny-Caffey syndrome, type 1	Epidermolytic hyperkeratosis	Ichthyosis histrix, Curth-Macklin type
Ichthyosis, cyclic, with epidermolytic hyperkeratosis	Keratosis palmoplantaris striata III;	Palmoplantar keratoderma
Epidermolytic hyperkeratosis	Ichthyosis with confetti	EpIchthyosis, cyclic, with epidermolytic hyperkeratosis

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Epilepsy, progressive myoclonic 2A (Lafora)	Epilepsy, progressive myoclonic 2B (Lafora)	Epilepsy, pyridoxine-dependent
Epilepsy, X-linked, with variable learning disabilities and behavior disorders	Intellectual developmental disorder, X-linked 50	Epileptic encephalopathy, early infantile, 12
Epileptic encephalopathy, early infantile, 2	Epileptic encephalopathy, early infantile, 3	Epileptic encephalopathy, early infantile, 8
Epileptic encephalopathy, early infantile, 9	Erythrocytosis, familial, 4	Erythrokeratodermia variabilis et progressiva 2
Möbius syndrome	Exudative vitreoretinopathy 1	Retinopathy of prematurity
Exudative vitreoretinopathy 2, X-linked	Norrie disease	Exudative vitreoretinopathy 4
Hyperostosis, endosteal	Osteopetrosis	Osteoporosis-pseudoglioma syndrome
Osteosclerosis	Polycystic liver disease 4 with or without kidney cysts	Fabry disease
LCHAD deficiency	Mitochondrial trifunctional protein deficiency	Galactokinase deficiency with cataracts
Galactose epimerase deficiency	Galactosemia	Galactosialidosis
Glutaric acidemia IIA	Glutaric acidemia IIB	Glutaric acidemia IIC
Glutaricaciduria, type I	Glycine N-methyltransferase deficiency	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a
Glycogen storage disease II	Gracile bone dysplasia	Kenny-Caffey syndrome, type 2
Hawkinsinuria	Tyrosinemia, type III	Hemochromatosis
Hemolytic anemia, G6PD deficient (favism)	Hennekam lymphangiectasia-lymphedema syndrome 1	HMG-CoA lyase deficiency

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Holocarboxylase synthetase deficiency	Homocystinuria due to MTHFR deficiency	Homocystinuria-megaloblastic anemia, cbl E type
Homocystinuria-megaloblastic anemia, cblG complementation type	Homocystinuria, B6-responsive and nonresponsive types	Thrombosis, hyperhomocysteinemic
Homocystinuria, cblD type, variant 1	Methylmalonic aciduria and homocystinuria, cblD type	Methylmalonic aciduria, cblD type, variant 2
HSD10 mitochondrial disease	Hypercalcemia, infantile, 1	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency
Methionine adenosyltransferase deficiency, automoso recessive	Hyperphenylalaninemia, BH4-deficient, A	Hyperphenylalaninemia, BH4-deficient, C
Hyperphenylalaninemia, BH4-deficient, D	Hypophosphatemic rickets	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
Ichthyosis bullosa of Siemens	Ichthyosis prematurity syndrome	Ichthyosis, congenital, 3
Ichthyosis, congenital, 11	Ichthyosis, congenital, 1	Ichthyosis, congenital, 10
Ichthyosis, congenital, 2	Ichthyosis, congenital, 5	Ichthyosis, congenital, 6
Ichthyosis, congenital, 8	Ichthyosis, congenital, 9	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
Ichthyosis, X-linked	Immunodeficiency 10	Myopathy, tubular aggregate, 1
Stormorken syndrome	Immunodeficiency 18, SCID variant	Immunodeficiency 19
Immunodeficiency 46	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	Isovaleric acidemia
Joubert syndrome 15	Joubert syndrome 8	Joubert syndrome with ocular defect Joubert syndrome-3
Leber congenital amaurosis 13	Leber congenital amaurosis 4	Retinitis pigmentosa, juvenile

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Cone-rod dystrophy	Leber congenital amaurosis 9	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis
Leigh syndrome	Leigh syndrome	Leigh syndrome
Leigh syndrome	Leigh syndrome	Leigh syndrome
Leigh syndrome due to mitochondrial complex I deficiency	Noonan syndrome 1	LEOPARD syndrome 1
Metachondromatosis	LIG4 syndrome	Maple syrup urine disease, type Ia
Maple syrup urine disease, type Ib	Maple syrup urine disease, type II	Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria, mut(0) typ	Methylmalonic aciduria, vitamin B12-responsive, cblA type	Methylmalonic aciduria, vitamin B12-responsive, cblB type
MHC class II deficiency, complementation group B	Microphthalmia, isolated, with coloboma 8	Microphthalmia, syndromic 9
microtubule-dependent vesicular transport, spindle assembly, and cell division. DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis.	Mucopolysaccharidosis VII	Multiple pterygium syndrome, lethal type
Myasthenic syndrome, congenital, 1A, slow-channel	Myasthenic syndrome, congenital, 1B, fast-channel	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
Neu-Laxova syndrome 1	Phosphoglycerate dehydrogenase deficiency	Neurofibromatosis, type 1
Neurofibromatosis, type 2	Noonan syndrome 4	Noonan syndrome 6
Noonan-like syndrome with loose anagen hair	NOR polyagglutination syndrome	Nystagmus 1, congenital
Nystagmus, infantile periodic alternating	Omenn syndrome	Severe combined immunodeficiency, Athabascan type
Opitz GBBB syndrome, type I	Optic disc anomalies with retinal and/or macular dystrophy	Lymphatic malformation 3

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Spastic paraplegia 44, autosomal recessive	Pelizaeus-Merzbacher-like due to GJC2 mutation	Phenylketonuria
Pitt-Hopkins syndrome	Polycystic kidney disease 4, with or without hepatic disease	Propionicacidemia
Renal tubular dysgenesis	Reticular dysgenesis	Retinitis pigmentosa 2
Retinitis pigmentosa 3	Macular degeneration, X-linked atrophic	Cone-rod dystrophy X-linked, 1
Retinitis pigmentosa 40	Night blindness, congenital stationary, autosomal dominant 2	Retinitis pigmentosa 43
Retinitis pigmentosa 59	Retinitis pigmentosa type 1	SCID, AR, T-negative/B-positive type
Seizures, benign neonatal, 2	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive
Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	Sialidosis, type I and type II	Sjogren-Larsson syndrome
Surfactant metabolism dysfunction, pulmonary, 1	Surfactant metabolism dysfunction, pulmonary, 2	T-cell immunodeficiency, congenital alopecia, and nail dystrophy
T-cell lymphopenia, infantile, with or without nail dystrophy	Thyroid dyshormonogenesis 1	Thyroid dyshormonogenesis 2A
Thyroid dyshormonogenesis 3	Thyroid dyshormonogenesis 6	Trifunctional protein deficiency
Tyrosinemia, type I	Tyrosinemia, type II	vascular smooth muscle cells; growth and differentiation factor that controls anterior/posterior patterning during embryonic development
VLCAD deficiency		