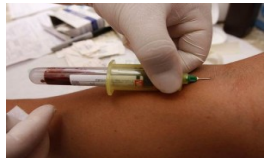
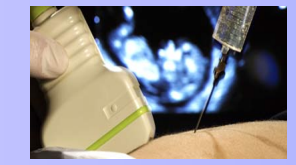
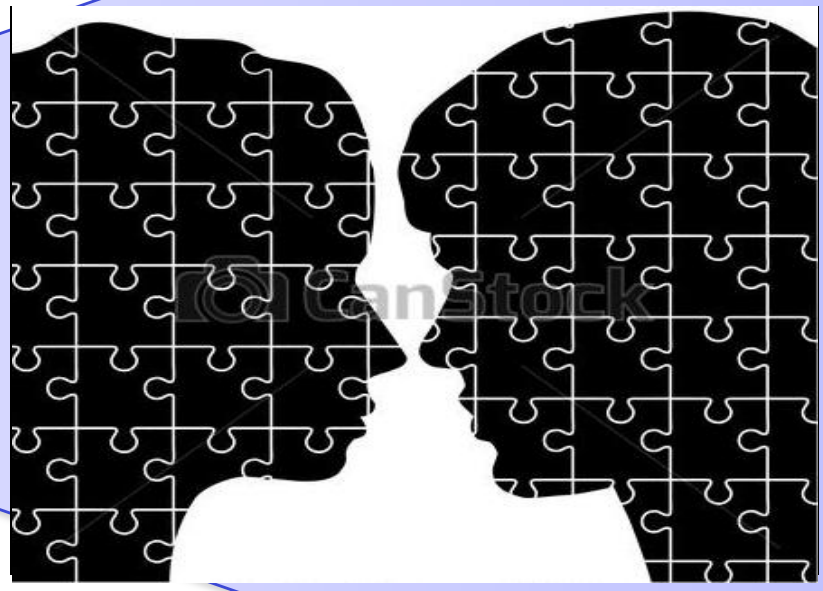
Questa presentazione sostituisce la lettura delle note informative allegate alla scheda che verrà sottoposta al paziente per accettazione. I punti essenziali sono indicati con:

TEST GENETICI PRENATALI





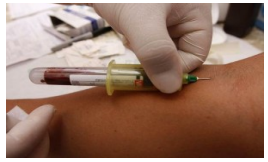


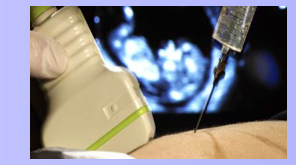
NON INVASIVI

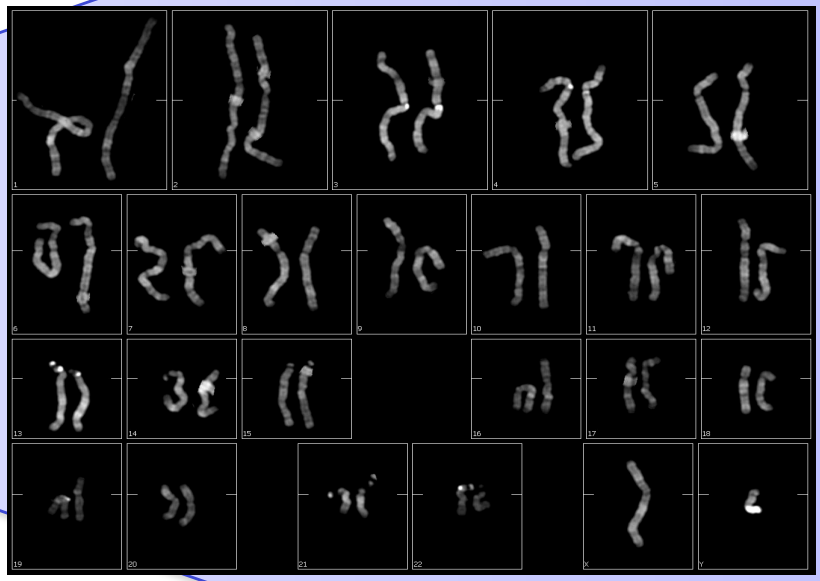
INVASIVI

Questa presentazione sostituisce la lettura delle note informative allegate alla scheda che verrà sottoposta al paziente per accettazione. I punti essenziali sono indicati con:

TEST GENETICI PRENATALI





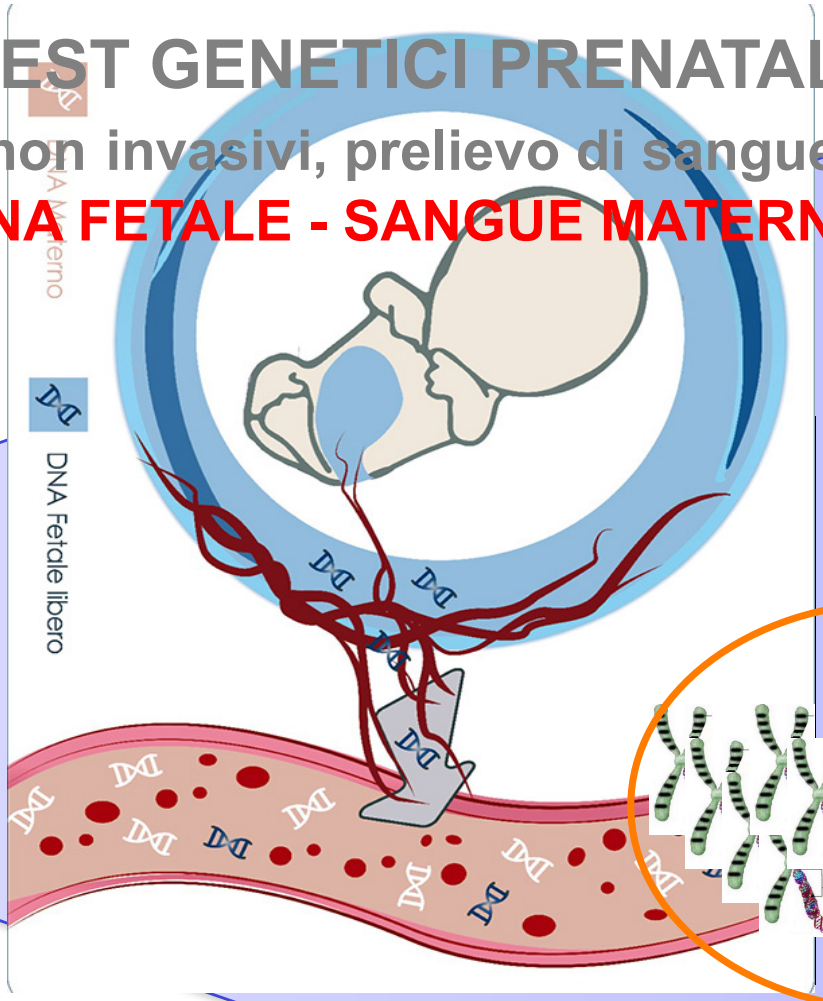


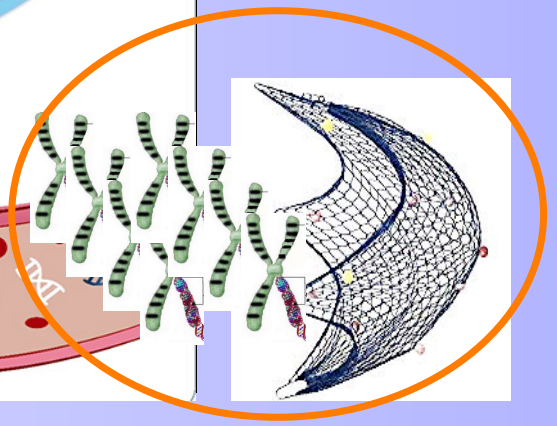
TEST GENETICI PRENATALI

non invasivi, prelievo di sangue

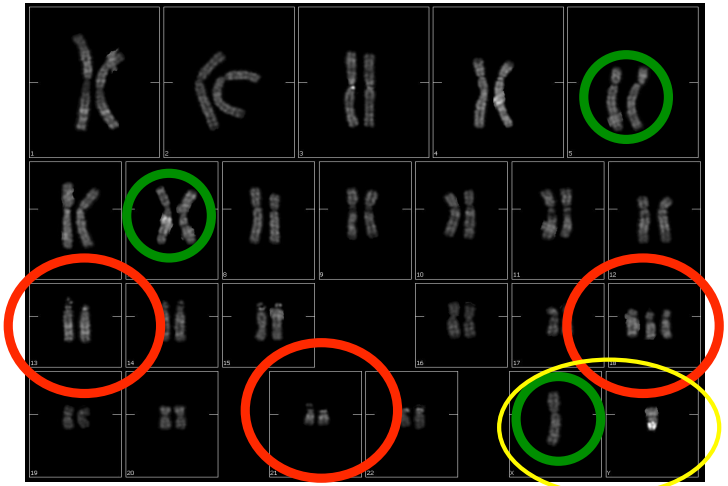
DNA FETALE - SANGUE MATERNO

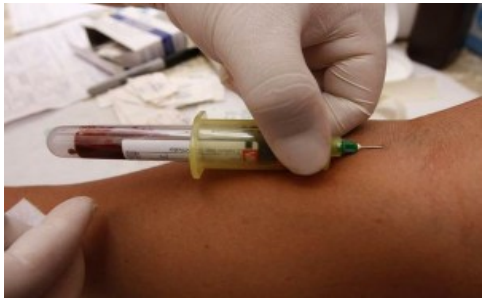
Fetal Fraction	Expected ratio for Trisomy
2-4%	1.02
10%	1.05
20%	1.10
40%	1.20





Test Prenatale Non Invasivo. Malattie genetiche: frequenti, gravi.

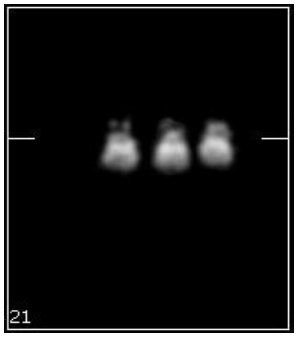




PrenatalSafe®

Harmony™ PRENATAL TEST

Ricerca su DNA fetale
Trisomie frequenti e gravi
Presenti dopo la 10^ settimana



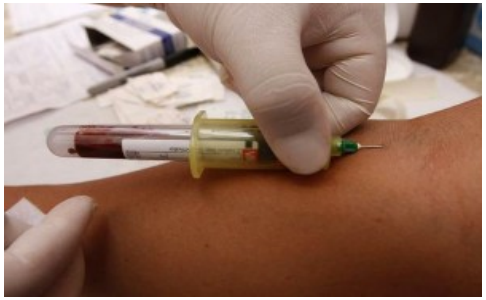
Sopravvivenza.

TRISOMIA 21	**1/750**	1a 88%	10a 82%
TRISOMIA 18	**1/2000**	1a 5%	1a 1%
TRISOMIA 13	**1/5000**	1a 10%	10a 0
XXX,X0,XXY,XYY	1/500		

Malattie genetiche frequenti e gravi:
Ricerca su DNA materno

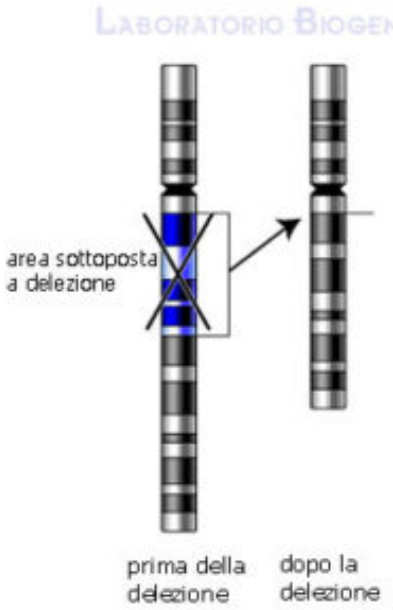
FIBROSI CISTICA	**1/2500**	40a 50%	
ATROFIA MUSCOLARE SPINALE	**1/10000**	SMA1 2a 5%	SMA2 20° 70% SMA3 adulta
RITARDO MENTALE FRAXA	**1/5000**	adulta	

DNA Fetale nel Plasma Materno

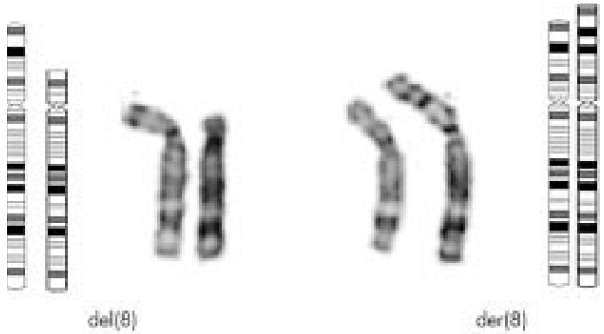


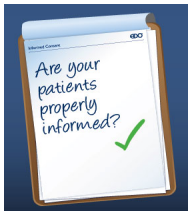
Anomalie Cromosomiche, trisomie e anomalie struttura
Cariotipo molecolare. 46 cromosomi

DELEZIONI	**0,30‰ (1/3000)**	1° 90%	10° >50%
DUPLICAZIONI	**0,15 ‰ (1/6000)**	1° 90%	10° >50%



PrenatalSafe®





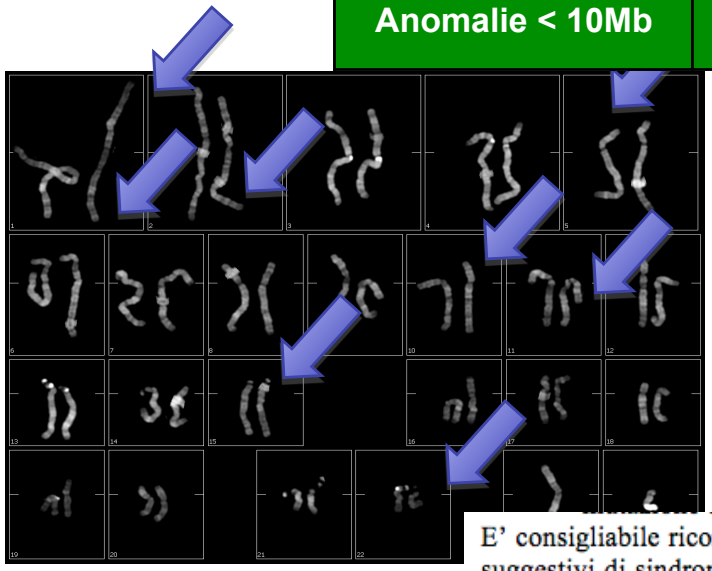
Prenatal screening

La analisi cffDNA delle microdelezioni (<10Mb) ricerca una serie di sindromi rare con una sensibilità clinica fra il 50 e il 95%.
L'analisi è consigliata come indagine di secondo livello in concomitanza con finding ecografici e consulenza genetica.

plus

PRENATAL EASY

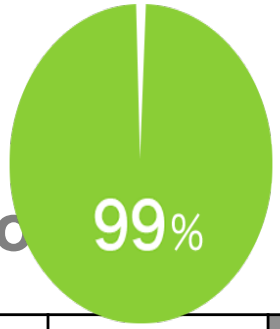
Casi di Anomalie Cromosomiche	Popolazione EU	‰ prevalence	% Anomalie Cromosomiche
Totale	10323	4,4	
Anomalie cromosomiche strutt.	1737	0,7	17 (40<10)
Anomalie < 10Mb			3

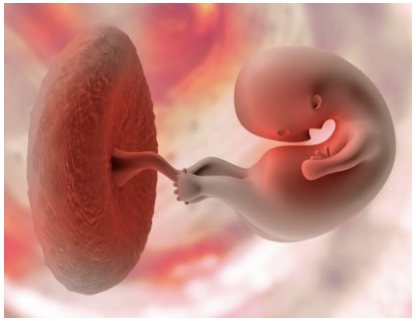


Sindrome da microdelezione	Regione cromosomica	Prevalenza (alla nascita)
Sindrome di DiGeorge	delezione 22q11.2	1/2.000 - 1/4.000
Sindrome Cri-du-chat	delezione 5p15.3	1/15.000 - 1/50.000
Sindrome di Prader-Willi	delezione 15q11.2	1/25.000
Sindrome Angelman	delezione 15q11.2	1/10.000 - 1/20.000
Sindrome da delezione 1p36	delezione 1p36	1/5.000 – 1/10.000
Sindrome di Wolf-Hirschhorn	delezione 4p16.3	1/20.000 -1/50.000
Sindrome di Jacobsen	delezione 11q23-q24.3	1/100.000
Sindrome di Langer-Giedion	delezione 8q24.11-q24.13	1/200.000
Sindrome di Smith-Magenis	delezione 17p11.2	1/15.000 - 1/25.000

E' consigliabile ricorrere all'utilizzo del **PrenatalSafe® Karyo Plus solo in determinati contesti clinici** (esempio dubbi ecografici suggestivi di sindrome da microdelezione cromosomica) per i quali risulta giustificato un approfondimento diagnostico di secondo livello.

NIPT
DNA fetale in sangue materno





Casi di Anomalie Cromosomiche	‰ prevalenza EU	Validazione clinica	cffDNA Sangue materno		cffDNA Sangue materno		DNA fetale placenta	Liquido Amniotico
Totale	4,4		SP	PPV	SN	NPV	Accuratezza%	Accuratezza %
T21 T18 T13	3,1	casi > 72000	99,9 99,9 99,9	99 99 82	99,5 99,9 99,9	99 99 99	99,96 99,87 99,92	100
XXX,XXY,XYY,X	0,6	casi > 72000	99,8-99,9	99-80	100 -98	100		

	SP	PPV	SE	NPV
Anomalie > 10Mb	0,4	casi > 72000	99,9	55,6	99-100	100
---	---	---	---	---	---	---

Falsi positivi > anomalia che non c'è
Falsi negativi > tutto bene ma non è vero

Ricerca anomalie fetali con cffDNA: performances

	Falsi +		Falsi -		Prev-popolaz /10000	Casi coorte
T21,18,13	12	1 6000	3	1 24000		72000
	Falsi +		**Falsi -**		**Sensibilità/ npv**	**Casi coorte**
Crom X e Y	**17**	1 4500	**1**	1 72000	99,99/100	72000
	Falsi +		**Falsi -**		**Sensibilità/ npv**	**Casi coorte**
Delezioni Duplicazioni	16	1 4500	1	1 72000	99,99/100	72000

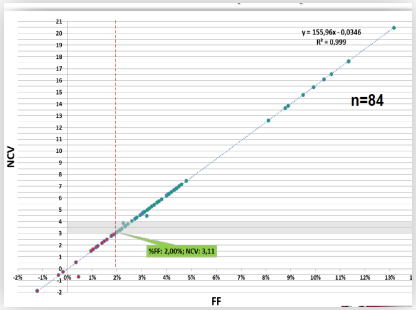
Falsi positivi > anomalia che non c'è

Falsi negativi > tutto bene ma non è vero

Ricerca anomalie cromosomi con cffDNA

PrenatalSafe® PRENATAL EASY

Trisomie & Cario. Analisi diretta del cariotipo molecolare fetale



DATI ANALISI

Risultato:

ANOMALIA	RISULTATI TEST
TRISOMIA 21	NON RILEVATO
TRISOMIA 18	NON RILEVATO
TRISOMIA 13	NON RILEVATO
ANEUPLOIDIE DEI CROMOSOMI SESSUALI XO, XXY, XYY, XXX	NON RILEVATO
ANEUPLOIDIE AUTOSOMICHE RARE	NON RILEVATO
DELEZIONI E DUPLICAZIONI (≥ 7 Mb)	NON RILEVATO
9 MICRODELEZIONI (< 7Mb) Vedi la sezione Note Tecniche	NON RILEVATO
SESSO FETALE	FEMMINA
FRAZIONE FETALE	12%

Interpretazione:

L'analisi su DNA libero circolante non ha rilevato alcuna anomalia tra quelle elencate nella tabella dei risultati.
Questo risultato è indicativo di un basso rischio per le condizioni analizzate.

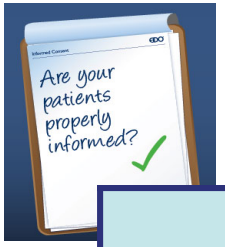
DATI ANALISI

Risultato:

ANOMALIA	RISULTATI TEST
TRISOMIA 21	NON RILEVATO
TRISOMIA 18	NON RILEVATO
TRISOMIA 13	NON RILEVATO
ANEUPLOIDIE DEI CROMOSOMI SESSUALI XO, XXY, XYY, XXX	NON RILEVATO
ANEUPLOIDIE AUTOSOMICHE RARE	NON RILEVATO
DELEZIONI E DUPLICAZIONI (≥ 7 Mb)	NON RILEVATO
9 MICRODELEZIONI (< 7Mb) Vedi la sezione Note Tecniche	RILEVATO (vedi sezione Interpretazione)
SESSO FETALE	MASCHIO
FRAZIONE FETALE	25%

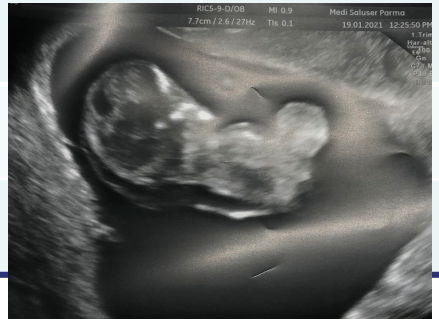
Interpretazione:

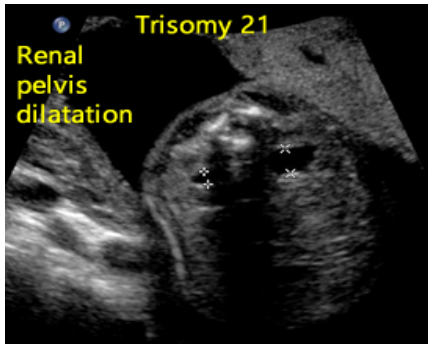
L'analisi ha rilevato un alto rischio di microdelezione nella regione 22q11.2, associata alla Sindrome da Delezione 22q11.2 anche nota come sindrome di DiGeorge (valore predittivo positivo stimato del 71.4%).
Non sono state rilevate altre anomalie tra quelle elencate nella tabella dei risultati (basso rischio).
Si consigliano consulenza genetica e diagnosi prenatale invasiva.

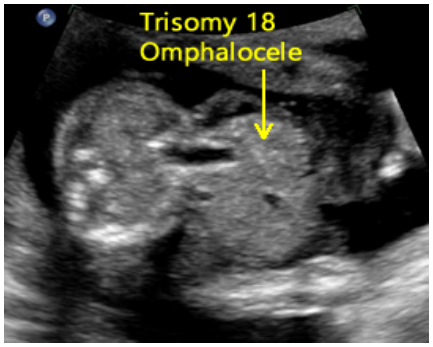


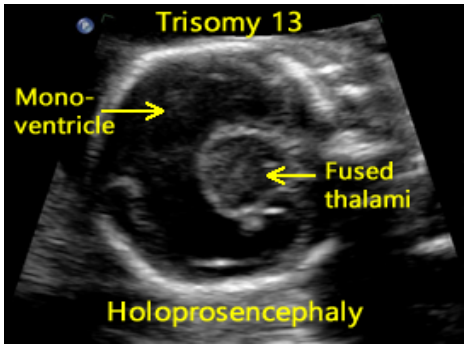
Ricerca trisomie fetali con cffDNA: performances

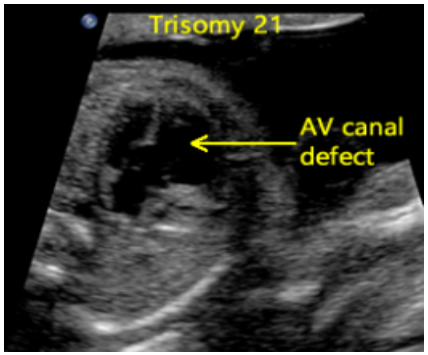
	Falsi +	Veri +	Falsi -	Prev-coorte /10000	Prev-popolaz /10000	Casi coorte
T21			**2**	120	15	72000
T18			**1**	30		72000
T13			**0**	13	0,6	

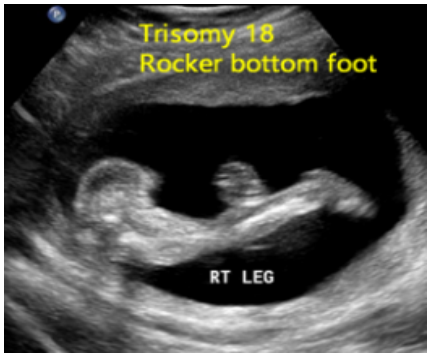


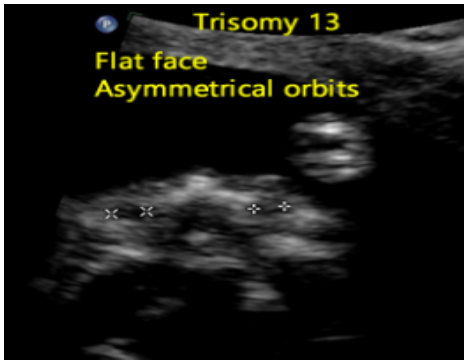


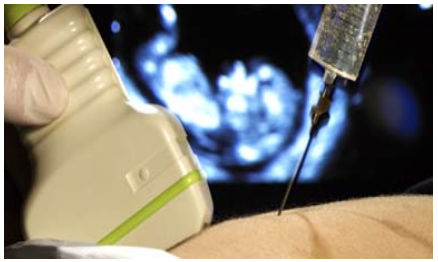








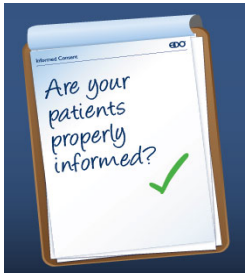




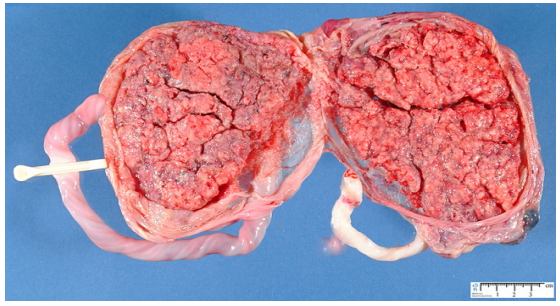
Fetal Aneuploidy Detection with cffDNA

Gravidanze multiple

DNA Fetale nel Plasma Materno



Twins/multiple	
2 max	Valore di rischio distribuito
regular	No sex, no identificazione feto affetto
IVF	No sex, no identificazione feto affetto
Oocytes etero	No sex, no identificazione feto affetto
Sensitivity T21*	93.7% *(FN/tested popul. 0.6%)*
Specificity T21*	99.8% *(FPR 0.23%)*
DNA fetal fraction	m 7,4% (12% single)
**Gil M et al 2015*	*Ultrasound Obstet Gynecol 45, 249*



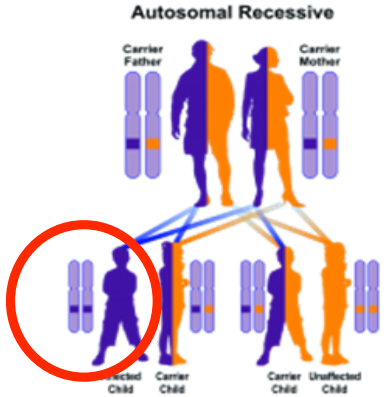
PluriGenTEST™

Test Prenatale Non Invasivo.
Malattie genetiche: frequenti, gravi.

Ricerca su DNA
di mamma/genitori > portatore sano.
Test diagnostico

Ricerca portatori malattie mendeliane ereditarie

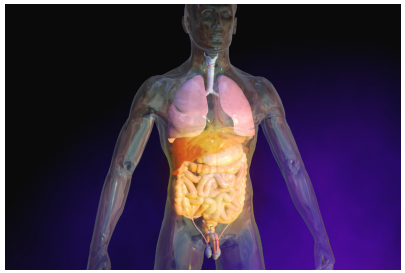
Fibrosi Cistica	port 1/25	malati 1 /2500	sensib. 90%
Ritardo mentale FRAXA	port. 1/260	1/1250M	99%
Atrofia Muscolare Spinale	port. 1/50	malati 1/10000	93%
	port. 1/40 ER	malati 1/6400	



Rischio residuo feto affetto

	FC	SMA
???? / ????	1/2500	1/10000
Neg 1 / ????	1/10000	1/100 000
Neg 1 / Neg 1	1/40000	1/1 000 000

FC
Fibrosi Cistica



SMA
Atrofia Muscolare Spinale

FRAXA
Ritardo Mentale

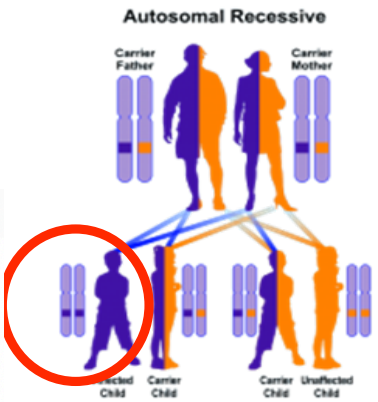
ritardo mentale: IQ tra 20 e 70

- deficit di memoria a breve termine di informazioni complesse
- ritardo nel linguaggio
- ridotte abilità visuo-spaziali
- ipersensibilità agli stimoli
- iperattività con deficit di attenzione
- comportamento autistico

- Macrocefalia con fronte, mento e orecchie sporgenti
- Macroorchidismo (<30ml) dopo la pubertà
- Anomalie connettivali: prolasso della mitrale, lassità articolare, piede piatto
- Disfunzioni ipotalamiche?

PluriGenTEST™

Test Prenatale Non Invasivo.
Malattie genetiche: 300 geni



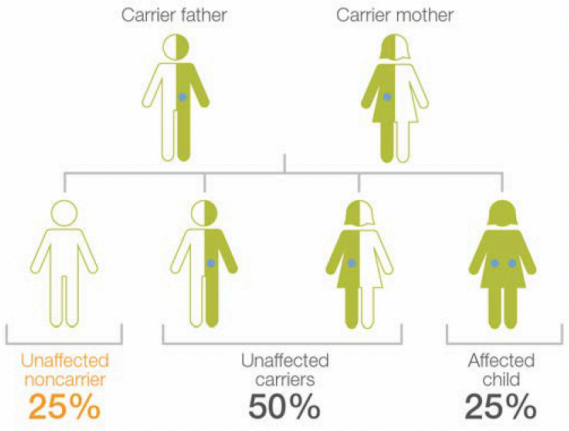
Ricerca su DNA di mamma/genitori > portatore sano. Test diagnostico

genescreen®

La genomica di nuova generazione

Negli ultimi anni, gli straordinari progressi conseguiti nel settore della genomica e delle biotecnologie hanno posto le basi per leggere e comprendere le informazioni contenute nel nostro DNA, il **genoma**. In particolare **le nuove tecnologie di sequenziamento, *Next Generation Sequencing* (NGS)**, ci permettono oggi di accedere alla sequenza del nostro DNA in modo più facile ed efficace, fornendo una valutazione approfondita dell'informazione genetica di ogni singolo individuo.

Ogni persona nasce, infatti, con caratteristiche genetiche che la differenziano dagli altri e che la rendono unica. Mentre la maggior parte delle differenze nella sequenza del DNA tra persone diverse è innocua, alcuni cambiamenti, definiti **mutazioni genetiche**, possono alterare la funzionalità genomica e rendere quella persona portatrice di una specifica malattia genetica trasmissibile ai propri figli.

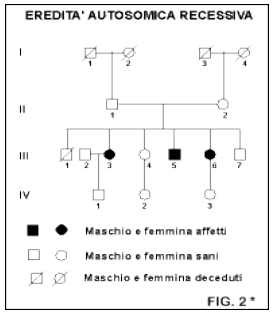


I portatori di malattie genetiche sono tipicamente individui sani, completamente privi di sintomi ed inconsapevoli di essere a rischio di trasmettere tale "errore" del DNA ai figli.

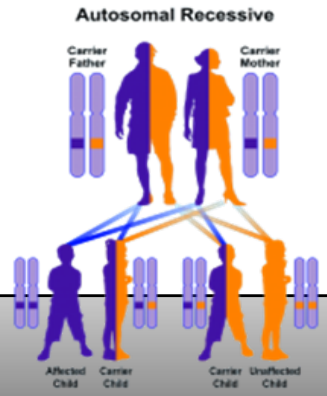
Il test GeneScreen® Easy

GeneScreen® Easy è un test diagnostico, sviluppato da GENOMA Group, che permette di eseguire un'analisi multipla di **oltre 600 malattie genetiche ereditarie**, tra cui quelle più frequenti nella popolazione italiana, come la Fibrosi Cistica, l'Anemia Falciforme, la Talassemia, la Sordità Ereditaria.

Test Prenatale Non Invasivo



PluriGenTEST™



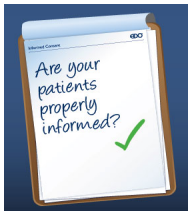
Ricerca portatori malattie mendeliane

Fibrosi Cistica	**90%** sensibilità (rischio residuo 1/95)
Atrofia muscolare spinale	**93%** sensibilità
Sordità NS	**90%** sensibilià (rischio residuo 1/350)
Ritardo mentale FRAXA	**99%** sensibilità

Probabilità feto affetto malattie mendeliane recessive

	Fibrosi Cistica	SMA
???? / ????	(1/25 x 1/25 x ¼) 1/2500	(1/50 x 1/50 x ¼) 1/10000
Neg 1 / ????	(1/25 x ¼ x 1/25 x ¼) 1/10000	(1/50 x 1/10 x 1/50 x ¼) 1/100 000
Neg 1 / Neg 1	(1/25 x ¼ x 1/25 x ¼ x ¼) 1/40000	(1/50 x 1/10 x 1/50 x 1/10 x ¼) 1/1 000 000
Etero / ???	(1 x 1/25 x ¼) 1/100	(1 x 1/50 x ¼) 1/200
Etero / Neg 1(75)	(1 x 1/25 x ¼x ¼) 1/400	Etero /Neg 1(93) (1 x 1/50 x 1/12 x ¼) 1/2400
Etero / Neg 2(90)	(1 x 1/25 x 1/10 x ¼) 1/1000	
Etero / Neg 3(95)	(1 x 1/25 x 1/20 x ¼) 1/2000	
Etero / Neg 4(99)	(1 x 1/25 x 1/100 x ¼) 1/10000	
Etero / Etero	¼	¼

Mutazioni: Neg 1(37), Neg3 (139-152), Neg4 (intero gene)



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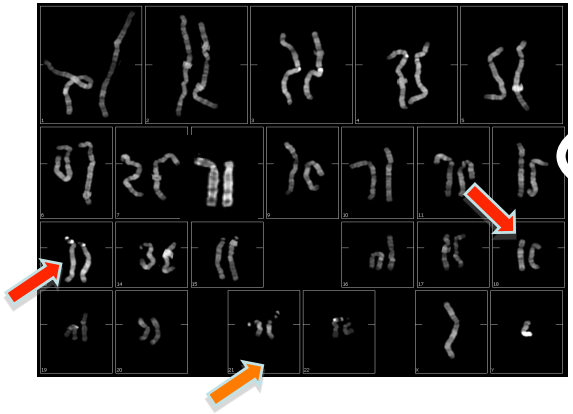
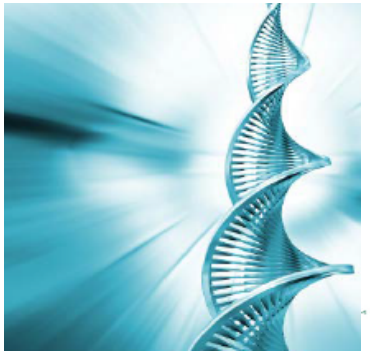


Anomalie	Anomalie ‰ nati	Specificità Sensibilità	Costo
Genetiche ereditarie	1,5	100 75-99	
Trisomie 21,18,13	3,1	99,7-99,9 98,4-99,5	650*
Cariotipo molecolare	4,1	99,9 (ppv 61) 99	
Microdelezioni 9 sindromi		- 50-95	
Altre malattie genetiche ereditarie SMA, sordità, talassemia	0,5	100 92-99	
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	



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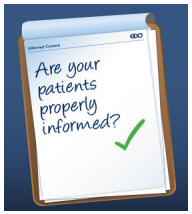

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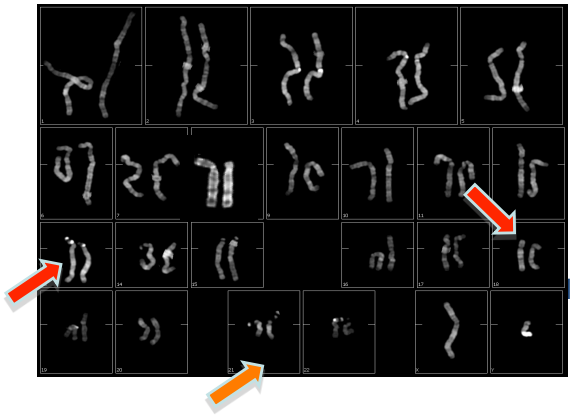
Microdelezioni 9 sindromi		- 50-95	
Altre malattie genetiche ereditarie sordità, talassemia	0,5	100 92-99	
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	

Fibrosi Cistica 30%
Ritardo mentale FRAXA 99%
Atrofia Muscolare Spinale 93%



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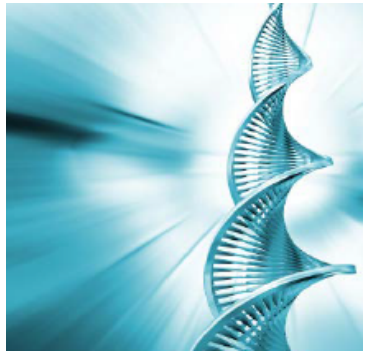

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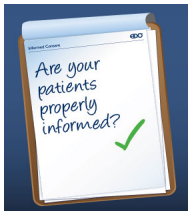

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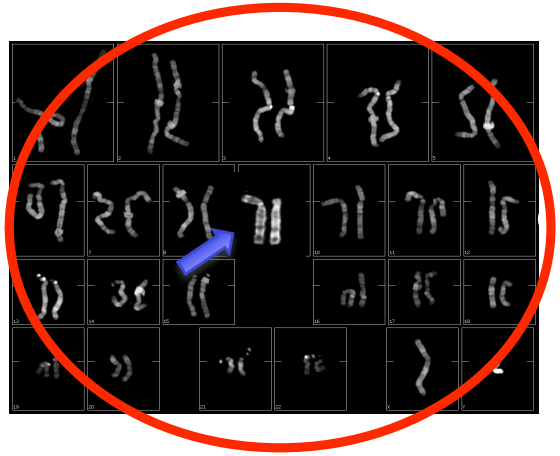

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Genetiche ereditarie	1,5	100 75-99	590
Trisomie 21,18,13	3,1	99,7-99,9 98,4-99,5	
Cariotipo molecolare comprese T21,18,13	4,1	99,9 (ppv 61) 99	895
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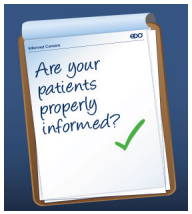

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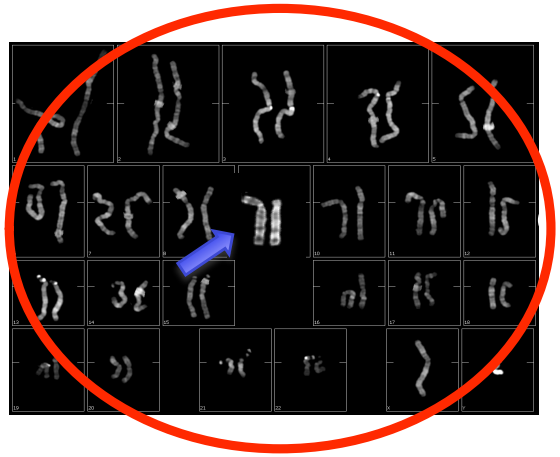
Microdelezioni 9 sindromi		- 50-95	
Altre malattie genetiche ereditarie sordità, talassemia,300 geni	2,5	100 92-99	
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	



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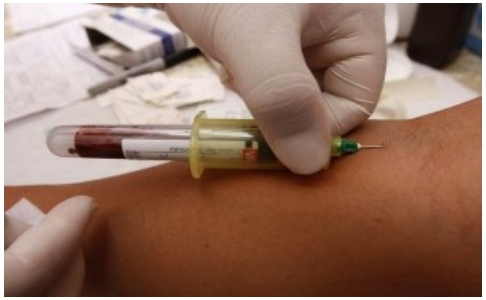
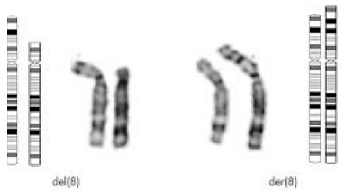
DNA Fetale nel Plasma Materno



Anomalie Cromosomiche, trisomie e anomalie struttura 46 cromosomi

Sopravvivenza. 1anno, 10 anni

DELEZIONI	**0,30‰ (1/3000)**	1° 90%	10° >50%
DUPLICAZIONI	**0,15 ‰ (1/6000)**	1° 90%	10° >50%



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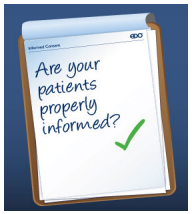

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Microdelezioni 9 sindromi		- 50-95	200
Altre malattie genetiche ereditarie Genescreen 300	2,5	100 92-99	780
Malattie genetiche Genesafe denovo	<0,01-0,4	>95 >70	780

Fibrosi Cistica 90%
Ritardo mentale FRAXA 99%
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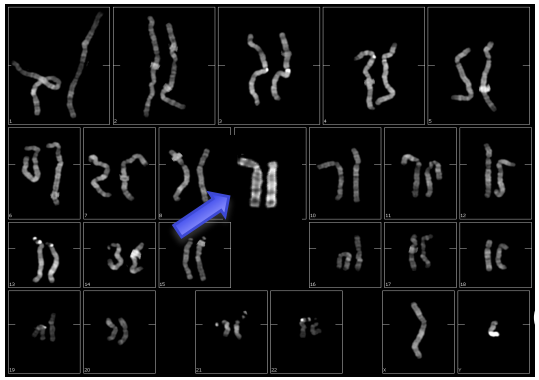

Sordità Neurosensoriale 90%

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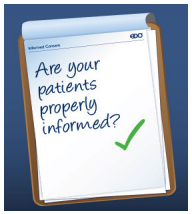

Prenatal screening

La analisi cffDNA delle microdelezioni (<10Mb) ricerca una serie di sindromi rare con una sensibilità clinica fra il 50 e il 95%.
L'analisi è consigliata come indagine di secondo livello in concomitanza con finding ecografici e consulenza genetica.

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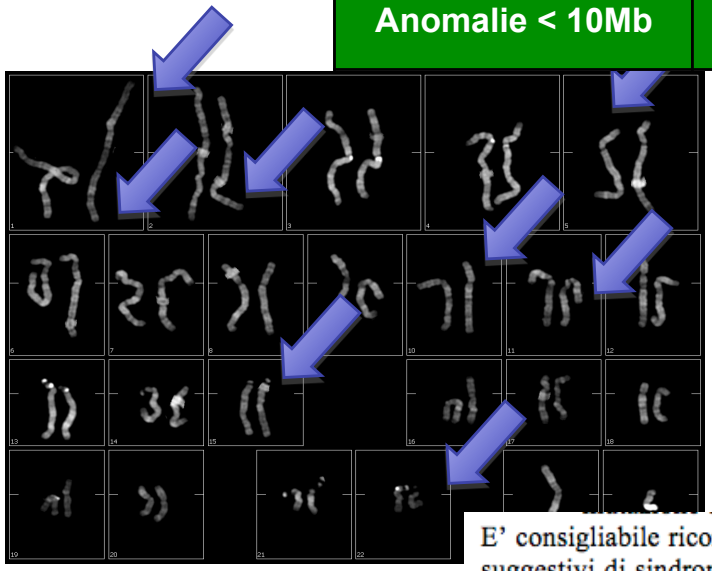
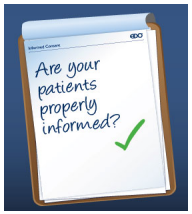
Casi di Anomalie Cromosomiche	Popolazione EU	‰ prevalence	% Anomalie Cromosomiche
Totale	10323	4,4	
Anomalie cromosomiche strutt.	1737	0,7	17 (40<10)
Anomalie < 10Mb			3



Sindrome da microdelezione	Regione cromosomica	Prevalenza (alla nascita)
Sindrome di DiGeorge	delezione 22q11.2	1/2.000 - 1/4.000
Sindrome Cri-du-chat	delezione 5p15.3	1/15.000 - 1/50.000
Sindrome di Prader-Willi	delezione 15q11.2	1/25.000
Sindrome Angelman	delezione 15q11.2	1/10.000 - 1/20.000
Sindrome da delezione 1p36	delezione 1p36	1/5.000 – 1/10.000
Sindrome di Wolf-Hirschhorn	delezione 4p16.3	1/20.000 -1/50.000
Sindrome di Jacobsen	delezione 11q23-q24.3	1/100.000
Sindrome di Langer-Giedion	delezione 8q24.11-q24.13	1/200.000
Sindrome di Smith-Magenis	delezione 17p11.2	1/15.000 - 1/25.000

E' consigliabile ricorrere all'utilizzo del **PrenatalSafe® Karyo Plus solo in determinati contesti clinici** (esempio dubbi ecografici suggestivi di sindrome da microdelezione cromosomica) per i quali risulta giustificato un approfondimento diagnostico di secondo livello.



Prenatal screening

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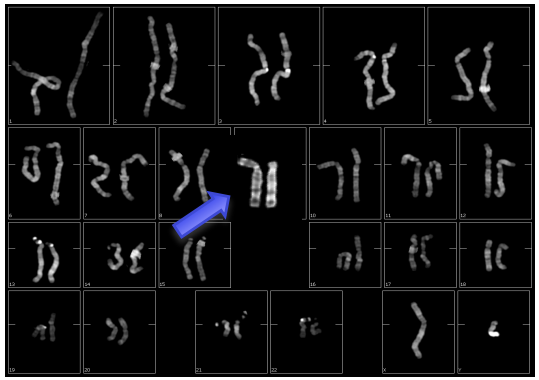

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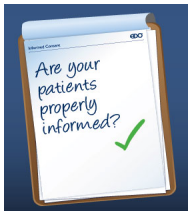

Prenatal screening

Malattie genetiche	Anomalie ‰	Specificità Sensibilità BIOLOGICA	Specificità Sensibilità TECNICA	Anomalie % tot
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	>95 >70	

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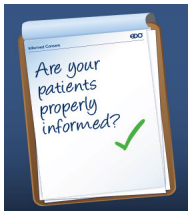
GENE	MALATTIE SINDROMICHE	
JAG1	Sindrome di Alagille	1/70000
CHD7	Sindrome di CHARGE	1/15000
HDAC8	Sindrome di Cornelia de Lange tipo 5	1/50000 Se 50%
NIPBL	Sindrome di Cornelia de Lange tipo 1	
MECP2	Sindrome di Rett	1/30000 Se bassa
NSD1	Sindrome di Sotos tipo1	
ASXL1	Sindrome di Bohring-Opitz	
SETBP1	Sindrome di Schinzel-Giedion	
	SINDROME DI NOONAN	
BRAF	Sindrome Cardio facio cutanea (CFS) tipo 1	
CBL	Sindrome di Noonan-simile con o senza leucemia mielomonocitica giovanile	1/2500 Se 50-75%
KRAS	Sindrome di Noonan /cancers	
MAP2K1	Sindrome Cardio facio cutanea (CFS) tipo e 3	
MAP2K2	Sindrome Cardio facio cutanea (CFS) tipo 4	
NRAS	Sindrome di Noonan 6/cancers	
PTPN11	Sindrome Noonan 1/ Sindrome di LEOPARD/cancers	
PTPN11	Leucemia mielomonocitica giovanile (JMML)	
RAF1	Sindrome di Noonan 5/Sindrome di LEOPARD 2	
RIT1	Sindrome di Noonan 8	
SHOC2	Sindrome Noonan-simile con capelli caduchi in fase anagen	
SOS1	Sindrome di Noonan 4	

GENE	PATOLOGIE SCHELETRICHE	
COL2A1	Acondrogenesi tipo 2	1/25000
FGFR3	Acondroplasia	
	Sindrome CATSHL	
	Sindromedi Crouzon con acanthosis nigricans	
	Ipocondroplasia	
	Sindrome di Muenke	
	Displasia tanatofora, tipo I	
	Displasia tanatofora, tipo II	
COL1A1	Sindrome di Ehlers-Danlos, classica	1/20000 Se 95%
	Sindrome di Ehlers-Danlos, tipo VIIA	
	Osteogenesi imperfetta, tipo I	
	Osteogenesi imperfetta, tipo IV	
	Osteogenesi imperfetta, tipo III	
	Osteogenesi imperfetta, tipo IV	
COL1A2	Sindrome di Ehlers-Danlos, forma cardiaco-valvolare	
	Sindrome di Ehlers-Danlos, tipo VIIB	
	Osteogenesi imperfetta, tipo II	
	Osteogenesi imperfetta, tipo III	
	Osteogenesi imperfetta, tipo IV	
	CRANIOSINOSTOSI	
FGFR2	Sindrome di Antley-Bixler senza anomalie genitali o disordini della steroidogenesi	1/2500 Se 20%
	Sindrome di Apert	
	Sindrome di Crouzon	
	Sindromedi Jackson-Weiss	
	Sindrome di Pfeiffer, tipo 1	
	Sindrome di Pfeiffer, tipo 2	
	Sindrome di Pfeiffer, tipo 3	



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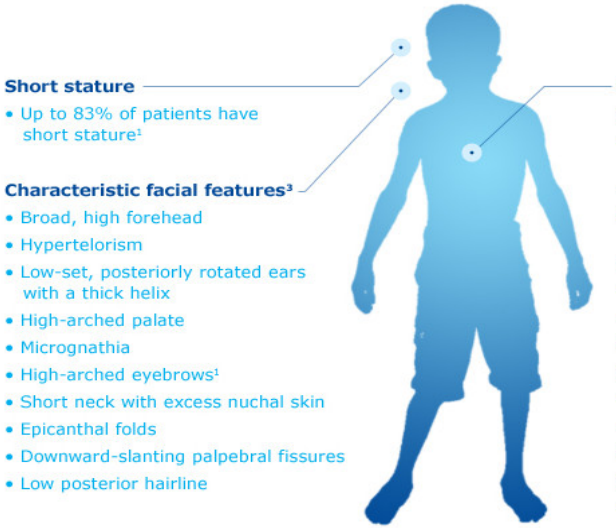
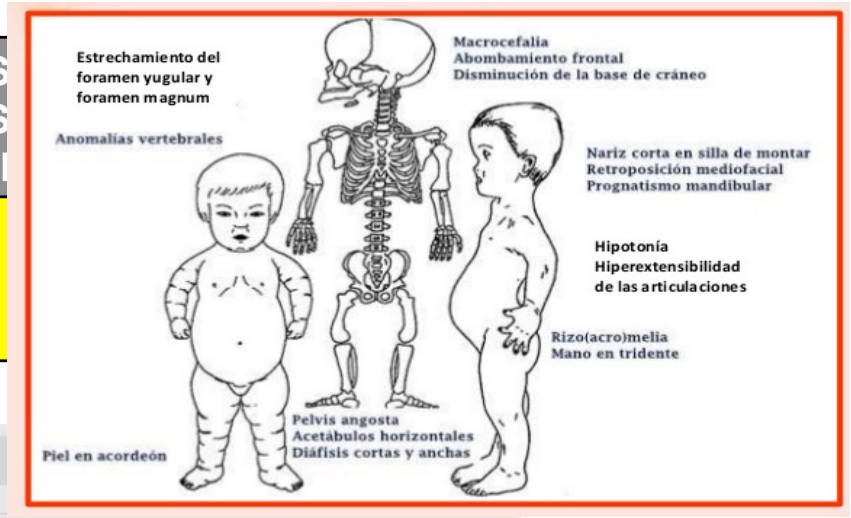
Malattie genetiche	Anomalie ‰	S S BI
	1-0,4	





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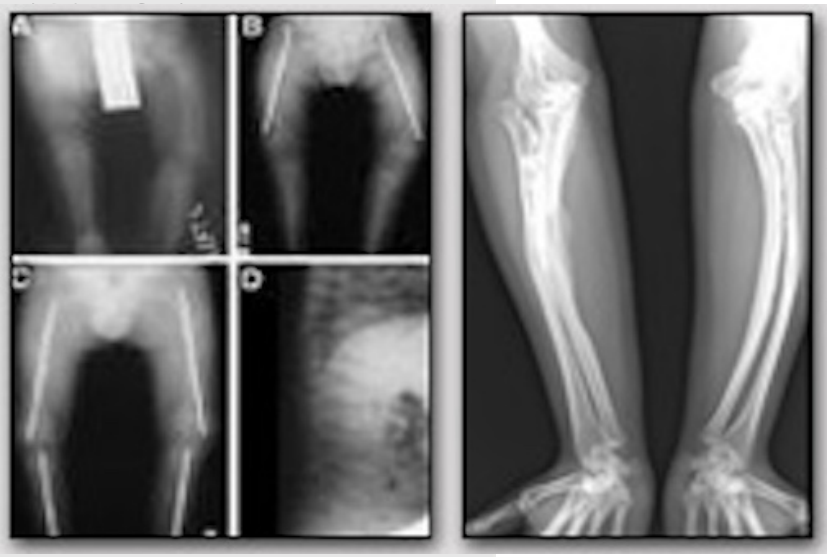
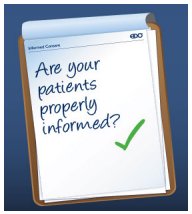
	Sindrome di Muenke
COL1A1	
COL1A2	
FGFR2	Sindrome di Pfeiffer, tipo 2
	Sindrome di Pfeiffer, tipo 3





Prenatal screening

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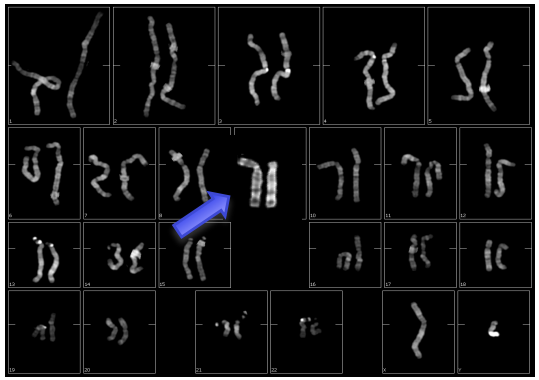

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Tabella 1: GeneScreen® Easy - Elenco dei geni analizzati e delle malattie genetiche investigate

Gene	OMIM Gene	Disease	OMIM Disease	Inheritance
ABCD1	300371	Adrenoleukodystrophy	300100	XLR
ABCC8	600509	Diabetes mellitus, noninsulin-dependent	125853	AD
		Diabetes mellitus, permanent neonatal	606176	AD, AR
		Diabetes mellitus, transient neonatal 2	610374	
		Hyperinsulinemic hypoglycemia, familial, 1	256450	AD, AR
		Hypoglycemia of infancy, leucine-sensitive	240800	AD
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, 2	605479	AR
		Cholestasis, progressive familial intrahepatic 2	601847	AR
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	225410	AR
EVC	604831	?Weyers acrofacial dysostosis	193530	AD
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of	201470	AR
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	AR
EVC2	607261	Ellis-van Creveld syndrome	225500	AR
ACAT1	607809	Alpha-methylacetoacetic aciduria	203750	AR
ADA	608958	Adenosine deaminase deficiency, partial	102700	SMo, AR
ACADVL	609575	VLCAD deficiency	201475	AR
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	264470	AR
AGL	610860	Glycogen storage disease IIIa	232400	AR
ACAD9	611103	Mitochondrial complex I deficiency, nuclear type 20	611126	AR
AGA	613228	Aspartylglucosaminuria	208400	AR
		Severe combined immunodeficiency due to ADA deficiency	102700	SMo, AR
		Glycogen storage disease IIIb	232400	AR
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	600121	AR
AGXT	604285	Hyperoxaluria, primary, type 1	259900	AR
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300	AD, AR
ALDH3A2	609523	Sjogren-Larsson syndrome	270200	AR
ALDOB	612724	Fructose intolerance, hereditary	229600	AR
ALG6	604566	Congenital disorder of glycosylation, type Ic	603147	AR

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ALMS1	606844	Alstrom syndrome	203800	AR
ALPL	171760	Hypophosphatasia, adult	146300	AD, AR
		Hypophosphatasia, childhood	241510	AR
		Hypophosphatasia, infantile	241500	AR
		Odontohypophosphatasia	146300	AD, AR
AMT	238310	Glycine encephalopathy	605899	AR
AR	313700	Androgen insensitivity	300068	XLR
		Androgen insensitivity, partial, with or without breast cancer	312300	XLR
		Hypospadias 1, X-linked	300633	XLR
		Spinal and bulbar muscular atrophy of Kennedy	313200	XLR
		Prostate cancer, susceptibility to	176807	AD, SMu
ARG1	608313	Argininemia	207800	AR
ARSA	607574	Metachromatic leukodystrophy	250100	AR
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200	AR
ARSE	300180	Chondrodysplasia punctata, X-linked recessive	302950	XLR
ASL	608310	Argininosuccinic aciduria	207900	AR
ASNS	108370	Asparagine synthetase deficiency	615574	AR
ASPA	608034	Canavan disease	271900	AR
ASS1	603470	Citrullinemia	215700	AR
ATM	607585	Ataxia-telangiectasia	208900	AR
		Lymphoma, B-cell non-Hodgkin, somatic		
		Lymphoma, mantle cell, somatic		
		T-cell prolymphocytic leukemia, somatic		
		Breast cancer, susceptibility to	114480	AD, SMu
ATP6V1B1	192132	Renal tubular acidosis with deafness	267300	AR
ATP7A	300011	Menkes disease	309400	XLR
		Occipital horn syndrome	304150	XLR
		Spinal muscular atrophy, distal, X-linked 3	300489	XLR
ATP7B	606882	Wilson disease	277900	AR
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic	300448	
		Alpha-thalassemia/mental retardation syndrome	301040	XLD

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		Mental retardation-hypotonic facies syndrome, X-linked	309580	XLR
BBS1	209901	Bardet-Biedl syndrome 1	209900	AR, DR
BBS10	610148	Bardet-Biedl syndrome 10	615987	AR
BBS12	610683	Bardet-Biedl syndrome 12	615989	AR
BBS2	606151	Bardet-Biedl syndrome 2	615981	AR
		Retinitis pigmentosa 74	616562	AR
BCHE	177400	Butyrylcholinesterase deficiency	617936	
		Apnea, postanesthetic, susceptibility to, due to BCHE deficiency	617936	
BCKDHA	608348	Maple syrup urine disease, type Ia	248600	AR
BCKDHB	248611	Maple syrup urine disease, type Ib	248600	AR
BCS1L	603647	Bjornstad syndrome	262000	AR
		GRACILE syndrome	603358	
		Leigh syndrome	256000	Mi, AR
		Mitochondrial complex III deficiency, nuclear type 1	124000	AR
BLM	604610	Bloom syndrome	210900	AR
BSND	606412	Bartter syndrome, type 4a	602522	AR
		Sensorineural deafness with mild renal dysfunction	602522	AR
BTD	609019	Biotinidase deficiency	253260	AR
BTK	300300	Agammaglobulinemia, X-linked 1	300755	XLR
		Isolated growth hormone deficiency, type III, with agammaglobulinemia	307200	XLR
CAPN3	114240	Muscular dystrophy, limb-girdle, autosomal dominant 4	618129	AD
		Muscular dystrophy, limb-girdle, autosomal recessive 1	253600	AR
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types	236200	AR
		Thrombosis, hyperhomocysteinemic	236200	AR
CC2D2A	612013	COACH syndrome	216360	AR
		Joubert syndrome 9	612285	AR
		Meckel syndrome 6	612284	AR
CD40LG	300386	Immunodeficiency, X-linked, with hyper-IgM	308230	XLR
CDH23	605516	Deafness, autosomal recessive 12	601386	AR
		Usher syndrome, type 1D	601067	AR, DR
		Usher syndrome, type 1D/F digenic	601067	AR, DR

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		Pituitary adenoma 5, multiple types	617540	AD
CEP290	610142	Bardet-Biedl syndrome 14	615991	AR
		Joubert syndrome 5	610188	AR
		Leber congenital amaurosis 10	611755	
		Meckel syndrome 4	611134	AR
		Senior-Loken syndrome 6	610189	AR
CERKL	608381	Retinitis pigmentosa 26	608380	
CFTR	602421	Congenital bilateral absence of vas deferens	277180	AR
		Cystic fibrosis	219700	AR
		Sweat chloride elevation without CF		
		Bronchiectasis with or without elevated sweat chloride 1, modifier of	211400	AD
		Hypertrypsinemia, neonatal		
		Pancreatitis, hereditary	167800	AD
CHM	300390	Choroideremia	303100	XLD
CHRNE	100725	Myasthenic syndrome, congenital, 4A, slow-channel	605809	AD, AR
		Myasthenic syndrome, congenital, 4B, fast-channel	616324	AR
		Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	608931	AR
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3	204200	AR
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5	256731	AR
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6	601780	AR
		Ceroid lipofuscinosis, neuronal, Kufs type, adult onset	204300	AR
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8	600143	AR
		Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	610003	AR
CLRN1	606397	Retinitis pigmentosa 61	614180	
		Usher syndrome, type 3A	276902	AR
CNGB3	605080	Achromatopsia 3	262300	AR
		Macular degeneration, juvenile	248200	AR
COL17A1	113811	Epidermolysis bullosa, junctional, localisata variant	226650	AR
		Epidermolysis bullosa, junctional, non-Herlitz type	226650	AR
		Epithelial recurrent erosion dystrophy	122400	AD
COL4A3	120070	Alport syndrome 2, autosomal recessive	203780	AR

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		Alport syndrome 3, autosomal dominant	104200	AD
		Hematuria, benign familial	141200	AD
COL4A4	120131	Alport syndrome 2, autosomal recessive	203780	AR
		Hematuria, familial benign	141200	AD
COL4A5	303630	Alport syndrome 1, X-linked	301050	XLD
COL7A1	120120	EBD inversa	226600	AR
		EBD, Bart type	132000	AD
		EBD, localisata variant		
		Epidermolysis bullosa dystrophica, AD	131750	AD
		Epidermolysis bullosa dystrophica, AR	226600	AR
		Epidermolysis bullosa pruriginosa	604129	AD, AR
		Epidermolysis bullosa, pretibial	131850	AD, AR
		Toenail dystrophy, isolated	607523	AD
		Transient bullous of the newborn	131705	AD, AR
CPS1	608307	Carbamoylphosphate synthetase I deficiency	237300	AR
		Pulmonary hypertension, neonatal, susceptibility to	615371	
		Venoocclusive disease after bone marrow transplantation		
CPT1A	600528	CPT deficiency, hepatic, type IA	255120	AR
CPT2	600650	CPT II deficiency, infantile	600649	AR
		CPT II deficiency, lethal neonatal	608836	AR
		CPT II deficiency, myopathic, stress-induced	255110	AD, AR
		Encephalopathy, acute, infection-induced, 4, susceptibility to	614212	AD, AR
CRB1	604210	Leber congenital amaurosis 8	613835	
		Pigmented paravenous chorioretinal atrophy	172870	AD
		Retinitis pigmentosa-12	600105	AR
CRTAP	605497	Osteogenesis imperfecta, type VII	610682	AR
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	254800	AR
CTNS	606272	Cystinosis, atypical nephropathic	219800	AR
		Cystinosis, late-onset juvenile or adolescent nephropathic	219900	AR
		Cystinosis, nephropathic	219800	AR
		Cystinosis, ocular nonnephropathic	219750	AR

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CTSD	116840	Ceroid lipofuscinosis, neuronal, 10	610127	AR
CTSK	601105	Pycnodysostosis	265800	AR
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	AR
		Aldosteronism, glucocorticoid-remediable	103900	AD
CYP11B2	124080	Aldosterone to renin ratio raised		
		Hypoaldosteronism, congenital, due to CMO I deficiency	203400	AR
		Hypoaldosteronism, congenital, due to CMO II deficiency	610600	AR
		Low renin hypertension, susceptibility to		
CYP17A1	609300	17,20-lyase deficiency, isolated	202110	AR
		17-alpha-hydroxylase/17,20-lyase deficiency	202110	AR
CYP19A1	107910	Aromatase deficiency	613546	
		Aromatase excess syndrome	139300	AD
CYP1B1	601771	Anterior segment dysgenesis 6, multiple subtypes	617315	
		Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	231300	AR
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	201910	AR
		Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	201910	AR
CYP27A1	606530	Cerebrotendinous xanthomatosis	213700	AR
DBT	248610	Maple syrup urine disease, type II	248600	AR
DCLRE1C	605988	Omenn syndrome	603554	AR
		Severe combined immunodeficiency, Athabascan type	602450	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	270400	AR
DHDDS	608172	Congenital disorder of glycosylation, type 1bb	613861	AR
		Developmental delay and seizures with or without movement abnormalities	617836	AD
		Retinitis pigmentosa 59	613861	AR
DKC1	300126	Dyskeratosis congenita, X-linked	305000	XLR
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	246900	AR
DMD	300377	Becker muscular dystrophy	300376	XLR
		Cardiomyopathy, dilated, 3B	302045	XL
		Duchenne muscular dystrophy	310200	XLR
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	608644	

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DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus	244400	AR
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	612444	
DOK7	610285	Fetal akinesia deformation sequence	208150	AR
		Myasthenic syndrome, congenital, 10	254300	AR
DPYD	612779	5-fluorouracil toxicity	274270	AR
		Dihydropyrimidine dehydrogenase deficiency	274270	AR
DYSF	603009	Miyoshi muscular dystrophy 1	254130	AR
		Muscular dystrophy, limb-girdle, autosomal recessive 2	253601	AR
		Myopathy, distal, with anterior tibial onset	606768	AR
EDA	300451	Ectodermal dysplasia 1, hypohidrotic, X-linked	305100	XLR
		Tooth agenesis, selective, X-linked 1	313500	XLD
EDAR	604095	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant	129490	AD
		Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	224900	AR
		Hair morphology 1, hair thickness	612630	
EIF2AK3	604032	Wolcott-Rallison syndrome	226980	AR
EIF2B1	606686	Leukoencephalopathy with vanishing white matter	603896	AR
EIF2B2	606454	Leukoencephalopathy with vanishing white matter	603896	AR
		Ovarioleukodystrophy	603896	AR
EIF2B3	606273	Leukoencephalopathy with vanishing white matter	603896	AR
EIF2B4	606687	Leukoencephalopathy with vanishing white matter	603896	AR
		Ovarioleukodystrophy	603896	AR
EIF2B5	603945	Leukoencephalopathy with vanishing white matter	603896	AR
		Ovarioleukodystrophy	603896	AR
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked	310300	XLR
ERCC6	609413	Cerebrooculofacioskeletal syndrome 1	214150	AR
		Cockayne syndrome, type B	133540	AR
		De Sanctis-Cacchione syndrome	278800	AR
		Premature ovarian failure 11	616946	AD
		UV-sensitive syndrome 1	600630	AR
		Lung cancer, susceptibility to	211980	AD, SMu
		Macular degeneration, age-related, susceptibility to, 5	613761	

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ERCC8	609412	Cockayne syndrome, type A	216400	AR
		UV-sensitive syndrome 2	614621	AR
ESCO2	609353	Roberts syndrome	268300	AR
		SC phocomelia syndrome	269000	AR
ETFA	608053	Glutaric acidemia IIA	231680	AR
ETFB	130410	Glutaric acidemia IIB	231680	AR
ETFDH	231675	Glutaric acidemia IIC	231680	AR
ETHE1	608451	Ethylmalonic encephalopathy	602473	AR
EVC2	607261	Ellis-van Creveld syndrome	225500	AR
		Weyers acrofacial dysostosis	193530	AD
EYS	612424	Retinitis pigmentosa 25	602772	AR
F11	264900	Factor XI deficiency, autosomal dominant	612416	
		Factor XI deficiency, autosomal recessive	612416	
F8	300841	Hemophilia A	306700	XLR
F9	300746	Hemophilia B	306900	XLR
		Thrombophilia, X-linked, due to factor IX defect	300807	
		Deep venous thrombosis, protection against	300807	
		Warfarin sensitivity	122700	AD
FAH	613871	Tyrosinemia, type I	276700	AR
FANCA	607139	Fanconi anemia, complementation group A	227650	AR
FANCC	613899	Fanconi anemia, complementation group C	227645	AR
FANCG	602956	Fanconi anemia, complementation group G	614082	
FH	136850	Fumarase deficiency	606812	AR
		Leiomyomatosis and renal cell cancer	150800	AD
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153	AR
		Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	606612	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	607155	AR
FKTN	607440	Cardiomyopathy, dilated, 1X	611615	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	253800	AR
		Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	613152	AR

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		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	611588	AR
FMR1	309550	Fragile X syndrome	300624	XLD
		Fragile X tremor/ataxia syndrome	300623	XLD
		Premature ovarian failure 1	311360	XL
G6PC	613742	Glycogen storage disease Ia	232200	AR
G6PD	305900	Hemolytic anemia, G6PD deficient (favism)	300908	XLD
		Resistance to malaria due to G6PD deficiency	611162	
GAA	606800	Glycogen storage disease II	232300	AR
GALC	606890	Krabbe disease	245200	AR
GALK1	604313	Galactokinase deficiency with cataracts	230200	AR
GALNS	612222	Mucopolysaccharidosis IVA	253000	AR
GALT	606999	Galactosemia	230400	AR
GAMT	601240	Cerebral creatine deficiency syndrome 2	612736	AR
GBA	606463	Gaucher disease, perinatal lethal	608013	AR
		Gaucher disease, type I	230800	AR
		Gaucher disease, type II	230900	AR
		Gaucher disease, type III	231000	AR
		Gaucher disease, type IIIC	231005	AR
		Lewy body dementia, susceptibility to	127750	AD
		Parkinson disease, late-onset, susceptibility to	168600	Mu, AD
GBE1	607839	Glycogen storage disease IV	232500	AR
		Polyglucosan body disease, adult form	263570	AR
GCDH	608801	Glutaricaciduria, type I	231670	AR
GFM1	606639	Combined oxidative phosphorylation deficiency 1	609060	AR
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	302800	XLD
GJB2	121011	Bart-Pumphrey syndrome	149200	AD
		Deafness, autosomal dominant 3A	601544	AD
		Deafness, autosomal recessive 1A	220290	AR
		Hystrix-like ichthyosis with deafness	602540	AD
		Keratitis-ichthyosis-deafness syndrome	148210	AD
		Keratoderma, palmoplantar, with deafness	148350	AD

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		Vohwinkel syndrome	124500	AD
GJB6	604418	Deafness, autosomal dominant 3B	612643	AD
		Deafness, autosomal recessive 1B	612645	AR
		Deafness, digenic GJB2/GJB6	220290	AR
		Ectodermal dysplasia 2, Clouston type	129500	AD
GLA	300644	Fabry disease	301500	XL
		Fabry disease, cardiac variant	301500	XL
GLB1	611458	GM1-gangliosidosis, type I	230500	AR
		GM1-gangliosidosis, type II	230600	AR
		GM1-gangliosidosis, type III	230650	AR
		Mucopolysaccharidosis type IVB (Morquio)	253010	AR
GLDC	238300	Glycine encephalopathy	605899	AR
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease	611890	AR
		Lethal congenital contracture syndrome 1	253310	AR
GNE	603824	Nonaka myopathy	605820	AR
		Sialuria	269921	AD
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	222765	AR
GNPTAB	607840	Mucolipidosis II alpha/beta	252500	AR
		Mucolipidosis III alpha/beta	252600	AR
GNS	607664	Mucopolysaccharidosis type IIID	252940	AR
GP9	173515	Bernard-Soulier syndrome, type C	231200	AR
GRHPR	604296	Hyperoxaluria, primary, type II	260000	AR
GUSB	611499	Mucopolysaccharidosis VII	253220	AR
HADHA	600890	Fatty liver, acute, of pregnancy	609016	AR
		HELLP syndrome, maternal, of pregnancy	609016	AR
		LCHAD deficiency	609016	AR
		Trifunctional protein deficiency	609015	AR
HADHB	143450	Trifunctional protein deficiency	609015	AR
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive	610738	AR
HBA1	141800	Erythrocytosis, 7	617981	
		Heinz body anemias, alpha-	140700	AD

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

		Hemoglobin H disease, nondeletional	613978	
		Methemoglobinemia, alpha type	617973	
		Thalassemias, alpha-	604131	
HBA2	141850	Erythrocytosis 7	617981	
		Heinz body anemia	140700	AD
		Hemoglobin H disease, deletional and nondeletional	613978	
		Thalassemia, alpha-	604131	
HBB	141900	Delta-beta thalassemia	141749	AD
		Erythrocytosis 6	617980	
		Heinz body anemia	140700	AD
		Hereditary persistence of fetal hemoglobin	141749	AD
		Methmoglobinemia, beta type	617971	
		Sickle cell anemia	603903	AR
		Thalassemia, beta	613985	
		Thalassemia-beta, dominant inclusion-body	603902	
		Malaria, resistance to	611162	
HEXA	606869	GM2-gangliosidosis, several forms	272800	AR
		Tay-Sachs disease	272800	AR
		Hex A pseudodeficiency	272800	AR
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	268800	AR
HFE	613609	Hemochromatosis	235200	AR
		Transferrin serum level QTL2	614193	
		Alzheimer disease, susceptibility to	104300	AD
		Microvascular complications of diabetes 7	612635	
		Porphyria cutanea tarda, susceptibility to	176100	AD, AR
		Porphyria variegata, susceptibility to	176200	AD
HFE2	608374	Hemochromatosis, type 2A	602390	AR
HGD	607474	Alkaptonuria	203500	AR
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930	AR
		Retinitis pigmentosa 73	616544	AR
HLCS	609018	Holocarboxylase synthetase deficiency	253270	AR

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HMGCL	613898	HMG-CoA lyase deficiency	246450	AR
HOGA1	613597	Hyperoxaluria, primary, type III	613616	
HPS1	604982	Hermansky-Pudlak syndrome 1	203300	AR
HPS3	606118	Hermansky-Pudlak syndrome 3	614072	AR
HSD17B4	601860	D-bifunctional protein deficiency	261515	AR
		Perrault syndrome 1	233400	AR
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	201810	AR
HYLS1	610693	Hydrolethalus syndrome	236680	AR
IDS	300823	Mucopolysaccharidosis II	309900	XLR
IDUA	252800	Mucopolysaccharidosis Ih	607014	AR
		Mucopolysaccharidosis Ih/s	607015	AR
		Mucopolysaccharidosis Is	607016	AR
IKBKAP	603722	Dysautonomia, familial	223900	AR
IL2RG	308380	Combined immunodeficiency, X-linked, moderate	312863	XLR
		Severe combined immunodeficiency, X-linked	300400	XLR
ISPD	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	614643	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	616052	AR
IVD	607036	Isovaleric acidemia	243500	AR
KCNJ11	600937	Diabetes mellitus, transient neonatal, 3	610582	AD
		Diabetes, permanent neonatal, with or without neurologic features	606176	AD, AR
		Hyperinsulinemic hypoglycemia, familial, 2	601820	AR
		Maturity-onset diabetes of the young, type 13	616329	AD
		Diabetes mellitus, type 2, susceptibility to	125853	AD
L1CAM	308840	Corpus callosum, partial agenesis of	304100	XLR
		CRASH syndrome	303350	XLR
		Hydrocephalus due to aqueductal stenosis	307000	XLR
		Hydrocephalus with congenital idiopathic intestinal pseudoobstruction	307000	XLR
		Hydrocephalus with Hirschsprung disease	307000	XLR

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		MASA syndrome	303350	XLR
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient	607855	AR
		Muscular dystrophy, limb-girdle, autosomal recessive 23	618138	AR
LAMA3	600805	Epidermolysis bullosa, generalized atrophic benign	226650	AR
		Epidermolysis bullosa, junctional, Herlitz type	226700	AR
		Laryngoonychocutaneous syndrome	245660	AR
LAMB3	150310	Amelogenesis imperfecta, type IA	104530	AD
		Epidermolysis bullosa, junctional, Herlitz type	226700	AR
		Epidermolysis bullosa, junctional, non-Herlitz type	226650	AR
LAMC2	150292	Epidermolysis bullosa, junctional, Herlitz type	226700	AR
		Epidermolysis bullosa, junctional, non-Herlitz type	226650	AR
LCA5	611408	Leber congenital amaurosis 5	604537	
LHCGR	152790	Leydig cell adenoma, somatic, with precocious puberty	176410	
		Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320	AR
		Leydig cell hypoplasia with pseudohermaphroditism	238320	AR
		Luteinizing hormone resistance, female	238320	AR
		Precocious puberty, male	176410	AD
LHX3	600577	Pituitary hormone deficiency, combined, 3	221750	AR
LIFR	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559	AR
LIPA	613497	Cholesteryl ester storage disease	278000	AR
		Wolman disease	278000	AR
LIPH	607365	Hypotrichosis 7	604379	AR
		Woolly hair, autosomal recessive 2 with or without hypotrichosis	604379	AR
LOXHD1	613072	Deafness, autosomal recessive 77	613079	AR
LPL	609708	Combined hyperlipidemia, familial	144250	AD
		Lipoprotein lipase deficiency	238600	AR
		High density lipoprotein cholesterol level QTL 11		
LRPPRC	607544	Leigh syndrome, French-Canadian type	220111	AR

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LYST	606897	Chediak-Higashi syndrome	214500	AR
MAN2B1	609458	Mannosidosis, alpha-, types I and II	248500	AR
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200	AR
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210	AR
MCOLN1	605248	Mucolipidosis IV	252650	AR
MECP2	300005	Encephalopathy, neonatal severe	300673	XLR
		Mental retardation, X-linked syndromic, Lubs type	300260	XLR
		Mental retardation, X-linked, syndromic 13	300055	XLR
		Rett syndrome	312750	XLD
		Rett syndrome, atypical	312750	XLD
		Rett syndrome, preserved speech variant	312750	XLD
		Autism susceptibility, X-linked 3	300496	XL
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	613668	AR
MEFV	608107	Familial Mediterranean fever, AD	134610	AD
		Familial Mediterranean fever, AR	249100	AR
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7	610951	AR
		Macular dystrophy with central cone involvement	616170	AR
MKS1	609883	Bardet-Biedl syndrome 13	615990	AR
		Joubert syndrome 28	617121	AR
		Meckel syndrome 1	249000	AR
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	604004	AR
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	251100	AR
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	AR
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	277400	AR
MMADHC	611935	Homocystinuria, cblD type, variant 1	277410	AR
MPI	154550	Congenital disorder of glycosylation, type Ib	602579	AR
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic	254450	
		Thrombocythemia 2	601977	SMu, AD

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		Thrombocytopenia, congenital amegakaryocytic	604498	AR
MPV17	137960	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	256810	AR
MTHFR	607093	Homocystinuria due to MTHFR deficiency	236250	AR
		Neural tube defects, susceptibility to	601634	AR
		Schizophrenia, susceptibility to	181500	AD
		Thromboembolism, susceptibility to	188050	AD
		Vascular disease, susceptibility to		
MTM1	300415	Myotubular myopathy, X-linked	310400	XLR
MTTP	157147	Abetalipoproteinemia	200100	AR
		Metabolic syndrome, protection against	605552	AD
MUSK	601296	Fetal akinesia deformation sequence	208150	AR
		Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	616325	AR
MUT	609058	Methylmalonic aciduria, mut(0) type	251000	AR
MYO7A	276903	Deafness, autosomal dominant 11	601317	AD
		Deafness, autosomal recessive 2	600060	AR
		Usher syndrome, type 1B	276900	AR
NAGLU	609701	Charcot-Marie-Tooth disease, axonal, type 2V	616491	AD
		Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	AR
NAGS	608300	N-acetylglutamate synthase deficiency	237310	AR
NBN	602667	Aplastic anemia	609135	
		Leukemia, acute lymphoblastic	613065	
		Nijmegen breakage syndrome	251260	AR
NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	618232	AR
NEB	161650	Nemaline myopathy 2, autosomal recessive	256030	AR
NPC1	607623	Niemann-Pick disease, type C1	257220	AR
		Niemann-Pick disease, type D	257220	AR
NPC2	601015	Niemann-pick disease, type C2	607625	AR
NPHP1	607100	Joubert syndrome 4	609583	AR
		Nephronophthisis 1, juvenile	256100	AR
		Senior-Loken syndrome-1	266900	AR
NPHS1	602716	Nephrotic syndrome, type 1	256300	AR

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NPHS2	604766	Nephrotic syndrome, type 2	600995	AR
NR2E3	604485	Enhanced S-cone syndrome	268100	AR
		Retinitis pigmentosa 37	611131	AD, AR
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	256800	AR
		Medullary thyroid carcinoma, familial	155240	AD
OCRL	300535	Dent disease 2	300555	XLR
		Lowe syndrome	309000	XLR
OPA3	606580	3-methylglutaconic aciduria, type III	258501	AR
		Optic atrophy 3 with cataract	165300	AD
OTC	300461	Ornithine transcarbamylase deficiency	311250	XLR
PAH	612349	Phenylketonuria	261600	AR
		Hyperphenylalaninemia, non-PKU mild	261600	AR
PANK2	606157	HARP syndrome	607236	AR
		Neurodegeneration with brain iron accumulation 1	234200	AR
PC	608786	Pyruvate carboxylase deficiency	266150	AR
PCCA	232000	Propionicacidemia	606054	AR
PCCB	232050	Propionicacidemia	606054	AR
PCDH15	605514	Deafness, autosomal recessive 23	609533	AR
		Usher syndrome, type 1D/F digenic	601067	DR, AR
		Usher syndrome, type 1F	602083	AR
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	312170	XLD
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	614111	
PEX1	602136	Heimler syndrome 1	234580	AR
		Peroxisome biogenesis disorder 1A (Zellweger)	214100	AR
		Peroxisome biogenesis disorder 1B (NALD/IRD)	601539	AR
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger)	614870	AR
		Peroxisome biogenesis disorder 6B	614871	AR
PEX12	601758	Peroxisome biogenesis disorder 3A (Zellweger)	614859	AR
		Peroxisome biogenesis disorder 3B	266510	AR
PEX2	170993	Peroxisome biogenesis disorder 5A (Zellweger)	614866	AR
		Peroxisome biogenesis disorder 5B	614867	AR

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PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger)	614872	AR
		Peroxisome biogenesis disorder 7B	614873	AR
PEX6	601498	Heimler syndrome 2	616617	AR
		Peroxisome biogenesis disorder 4A (Zellweger)	614862	AR
		Peroxisome biogenesis disorder 4B	614863	AD, AR
PEX7	601757	Peroxisome biogenesis disorder 9B	614879	AR
		Rhizomelic chondrodysplasia punctata, type 1	215100	AR
PFKM	610681	Glycogen storage disease VII	232800	AR
PHGDH	606879	Neu-Laxova syndrome 1	256520	AR
		Phosphoglycerate dehydrogenase deficiency	601815	AR
PKHD1	606702	Polycystic kidney disease 4, with or without hepatic disease	263200	AR
PLA2G6	603604	Infantile neuroaxonal dystrophy 1	256600	AR
		Neurodegeneration with brain iron accumulation 2B	610217	AR
		Parkinson disease 14, autosomal recessive	612953	AR
PMM2	601785	Congenital disorder of glycosylation, type Ia	212065	AR
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type)	203700	AR
		Mitochondrial DNA depletion syndrome 4B (MNGIE type)	613662	AR
		Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	607459	AR
		Progressive external ophthalmoplegia, autosomal dominant 1	157640	AD
		Progressive external ophthalmoplegia, autosomal recessive 1	258450	AR
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	253280	AR
		Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	613151	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	613157	AR
		Retinitis pigmentosa 76	617123	AR
POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	236670	AR

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		Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	613155	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	609308	AR
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	613150	AR
		Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	613156	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	613158	AR
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1	256730	AR
PROP1	601538	Pituitary hormone deficiency, combined, 2	262600	AR
PRPS1	311850	Arts syndrome	301835	XLR
		Charcot-Marie-Tooth disease, X-linked recessive, 5	311070	XLR
		Deafness, X-linked 1	304500	XL
		Gout, PRPS-related	300661	XLR
		Phosphoribosylpyrophosphate synthetase superactivity	300661	XLR
PSAP	176801	Combined SAP deficiency	611721	AR
		Gaucher disease, atypical	610539	
		Krabbe disease, atypical	611722	AR
		Metachromatic leukodystrophy due to SAP-b deficiency	249900	AR
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	261640	AR
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1	600462	AR
PYGM	608455	McArdle disease	232600	AR
RAB23	606144	Carpenter syndrome	201000	AR
RAG1	179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity	609889	
		Combined cellular and humoral immune defects with granulomas	233650	AR
		Omenn syndrome	603554	AR
		Severe combined immunodeficiency, B cell-negative	601457	AR

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RAG2	179616	Combined cellular and humoral immune defects with granulomas	233650	AR
		Omenn syndrome	603554	AR
		Severe combined immunodeficiency, B cell-negative	601457	AR
RAPSN	601592	Fetal akinesia deformation sequence	208150	AR
		Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	616326	AR
RARS2	611524	Pontocerebellar hypoplasia, type 6	611523	AR
RDH12	608830	Leber congenital amaurosis 13	612712	AD, AR
RMRP	157660	Anauxetic dysplasia 1	607095	AR
		Cartilage-hair hypoplasia	250250	AR
		Metaphyseal dysplasia without hypotrichosis	250460	AR
RPE65	180069	Leber congenital amaurosis 2	204100	AR
		Retinitis pigmentosa 20	613794	AR
RPGRIP1L	610937	COACH syndrome	216360	AR
		Joubert syndrome 7	611560	AR
		Meckel syndrome 5	611561	AR
RS1	300839	Retinoschisis	312700	XLR
SACS	604490	Spastic ataxia, Charlevoix-Saguenay type	270550	AR
SAMHD1	606754	Chilblain lupus 2	614415	AD
		Aicardi-Goutieres syndrome 5	612952	AR
SBDS	607444	Shwachman-Diamond syndrome	260400	AR
		Aplastic anemia, susceptibility to	609135	
SEPSECS	613009	Pontocerebellar hypoplasia type 2D	613811	AR
SERPINA1	107400	Emphysema due to AAT deficiency	613490	AR
		Emphysema-cirrhosis, due to AAT deficiency	613490	AR
		Hemorrhagic diathesis due to antithrombin Pittsburgh	613490	AR
		Pulmonary disease, chronic obstructive, susceptibility to	606963	
SGCA	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3	608099	AR
SGCB	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4	604286	AR
SGCG	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5	253700	AR
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A)	252900	AR

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SLC12A3	600968	Gitelman syndrome	263800	AR
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	218000	AR
SLC17A5	604322	Salla disease	604369	AR
		Sialic acid storage disorder, infantile	269920	AR
SLC22A5	603377	Carnitine deficiency, systemic primary	212140	AR
SLC25A13	603859	Citrullinemia, adult-onset type II	603471	AR
		Citrullinemia, type II, neonatal-onset	605814	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970	AR
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	212138	AR
SLC26A2	606718	Achondrogenesis Ib	600972	AR
		Atelosteogenesis, type II	256050	AR
		De la Chapelle dysplasia	256050	AR
		Diastrophic dysplasia	222600	AR
		Diastrophic dysplasia, broad bone-platyspondylic variant	222600	AR
		Epiphyseal dysplasia, multiple, 4	226900	AR
SLC26A4	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791	AR
		Pendred syndrome	274600	AR
SLC37A4	602671	Glycogen storage disease Ib	232220	AR
		Glycogen storage disease Ic	232240	AR
SLC39A4	607059	Acrodermatitis enteropathica	201100	AR
SLC4A11	610206	Corneal dystrophy, Fuchs endothelial, 4	613268	
		Corneal endothelial dystrophy and perceptive deafness	217400	AR
		Corneal endothelial dystrophy, autosomal recessive	217700	AR
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	300352	XLR
SMN1	600354	Spinal muscular atrophy-1	253300	AR
		Spinal muscular atrophy-2	253550	AR
		Spinal muscular atrophy-3	253400	AR
		Spinal muscular atrophy-4	271150	AR
SMPD1	607608	Niemann-Pick disease, type A	257200	AR
STAR	600617	Lipoid adrenal hyperplasia	201710	AR

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SUMF1	607939	Multiple sulfatase deficiency	272200	AR
TAT	613018	Tyrosinemia, type II	276600	AR
TCIRG1	604592	Osteopetrosis, autosomal recessive 1	259700	AR
TFR2	604720	Hemochromatosis, type 3	604250	AR
TGM1	190195	Ichthyosis, congenital, autosomal recessive 1	242300	AR
TH	191290	Segawa syndrome, recessive	605407	AR
TMEM216	613277	Joubert syndrome 2	608091	AR
		Meckel syndrome 2	603194	AR
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2	204500	AR
		Spinocerebellar ataxia, autosomal recessive 7	609270	AR
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive	225750	AD, AR
		Chilblain lupus	610448	AD
		Vasculopathy, retinal, with cerebral leukodystrophy	192315	AD
		Systemic lupus erythematosus, susceptibility to	152700	AD
TRIM37	605073	Mulibrey nanism	253250	AR
TSEN2	608753	Pontocerebellar hypoplasia type 2B	612389	AR
TSEN34	608754	Pontocerebellar hypoplasia type 2C	612390	
TSEN54	608755	Pontocerebellar hypoplasia type 5	610204	AR
		Pontocerebellar hypoplasia type 2A	277470	AR
		Pontocerebellar hypoplasia type 4	225753	AR
TSFM	604723	Combined oxidative phosphorylation deficiency 3	610505	AR
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4	275100	AR
TTC37	614589	Trichohepatoenteric syndrome 1	222470	AR
TTPA	600415	Ataxia with isolated vitamin E deficiency	277460	AR
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041	AR
TYR	606933	Albinism, oculocutaneous, type IA	203100	AR
		Albinism, oculocutaneous, type IB	606952	
		Waardenburg syndrome/albinism, digenic	103470	AD
		Skin/hair/eye pigmentation 3, blue/green eyes	601800	
		Skin/hair/eye pigmentation 3, light/dark/freckling skin	601800	
		Melanoma, cutaneous malignant, susceptibility to, 8	601800	

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UGT1A1	191740	Crigler-Najjar syndrome, type I	218800	AR
		Crigler-Najjar syndrome, type II	606785	AR
		Hyperbilirubinemia, familial transient neonatal	237900	AR
		Bilirubin, serum level of, QTL1	601816	
		Gilbert syndrome	143500	AR
USH1C	605242	Deafness, autosomal recessive 18A	602092	AR
		Usher syndrome, type 1C	276904	AR
USH2A	608400	Retinitis pigmentosa 39	613809	
		Usher syndrome, type 2A	276901	AR
VPS13A	605978	Choreoacanthocytosis	200150	AR
VPS13B	607817	Cohen syndrome	216550	AR
VRK1	602168	Pontocerebellar hypoplasia type 1A	607596	AR
WAS	300392	Neutropenia, severe congenital, X-linked	300299	XLR
		Thrombocytopenia, X-linked	313900	XLR
		Thrombocytopenia, X-linked, intermittent	313900	XLR
		Wiskott-Aldrich syndrome	301000	XLR
WNT10A	606268	Odontoonychodermal dysplasia	257980	AR
		Schopf-Schulz-Passarge syndrome	224750	AR
		Tooth agenesis, selective, 4	150400	AD, AR
XPA	611153	Xeroderma pigmentosum, group A	278700	AR
XPC	613208	Xeroderma pigmentosum, group C	278720	AR
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive	270700	AR

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