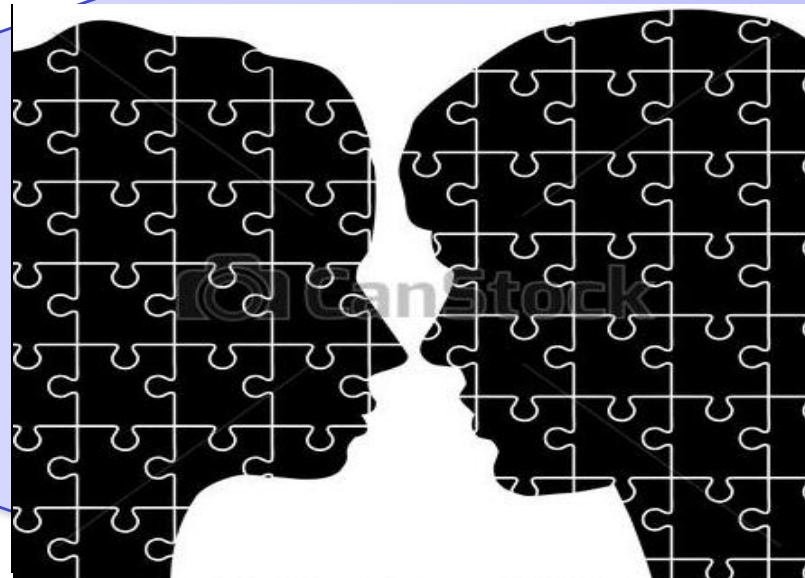


Questa presentazione sostituisce la lettura delle note informative allegate alla scheda che verrà sottoposta al paziente per accettazione. I punti essenziali sono indicati con:



## TEST GENETICI PRENATALI



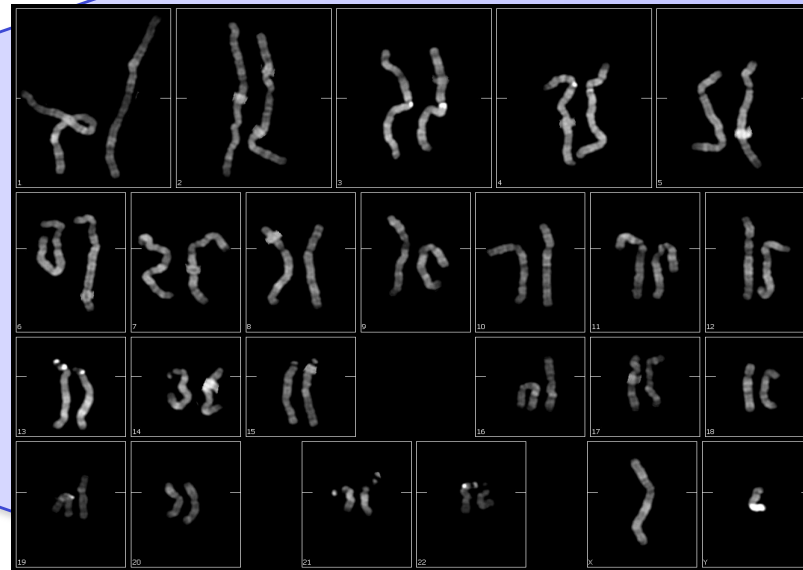
NON INVASIVI

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:

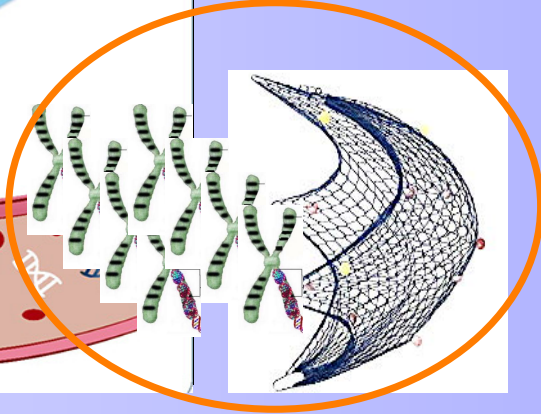
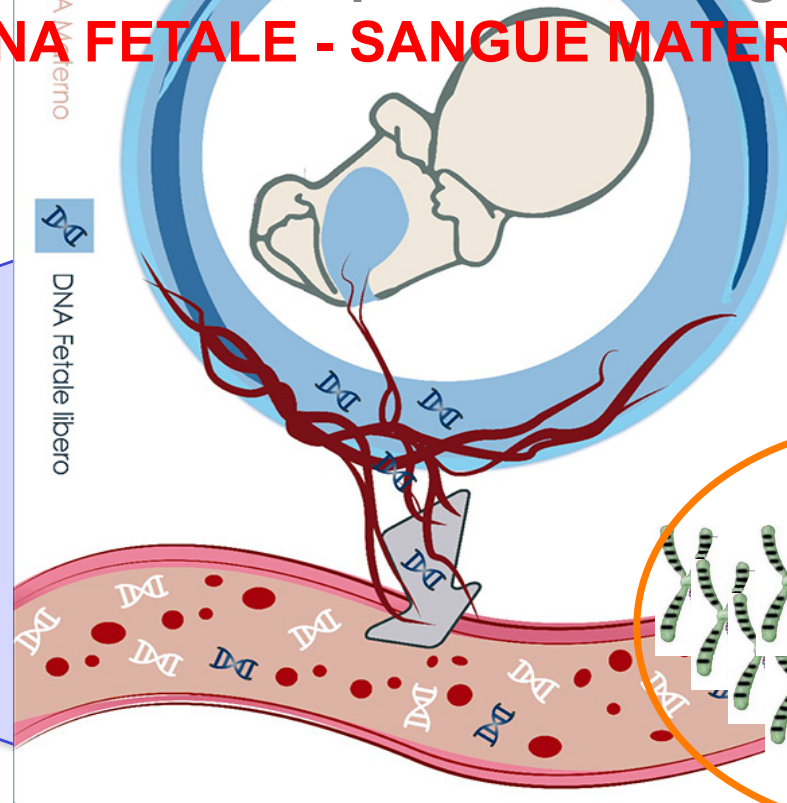


# TEST GENETICI PRENATALI



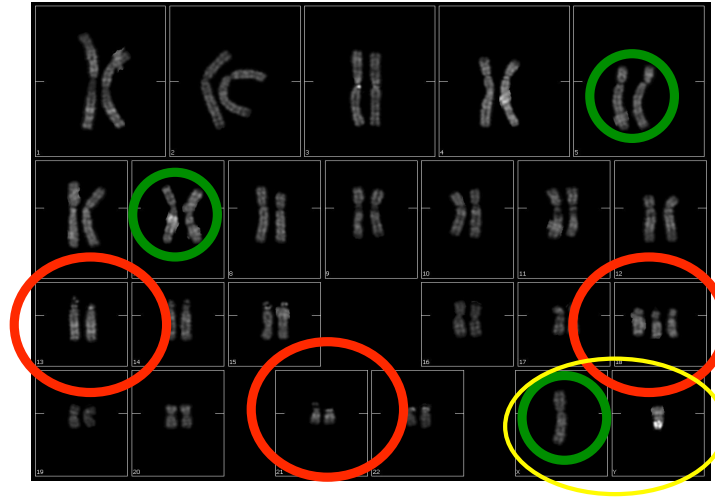
# TEST GENETICI PRENATALI non invasivi, prelievo di sangue **DNA FETALE - SANGUE MATERNO**

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

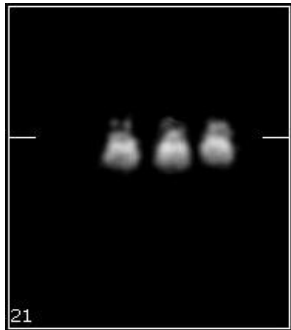


Property of  
Lamberto Camurri Ph.D.

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



**Ricerca su DNA fetale**  
Trisomie frequenti e gravi  
Presenti dopo la 10<sup>a</sup> settimana



Sopravvivenza.

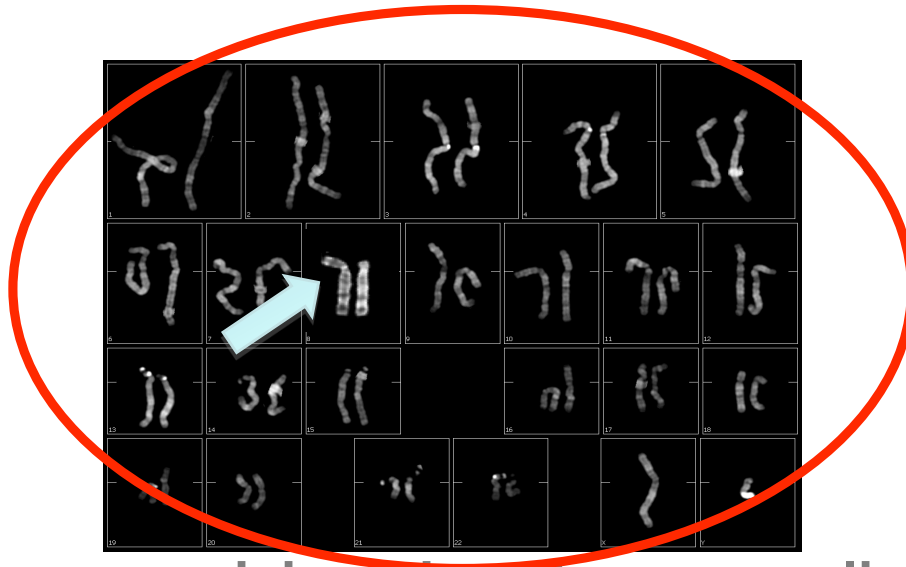
<b>TRISOMIA 21</b>	<b>1/750</b>	1a 88%	10a 82%
<b>TRISOMIA 18</b>	<b>1/2000</b>	1a 5%	1a 1%
<b>TRISOMIA 13</b>	<b>1/5000</b>	1a 10%	10a 0
XXX,X0,XXY,XXY	1/500		

**Malattie genetiche frequenti e gravi:  
Ricerca su DNA  
materno**

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

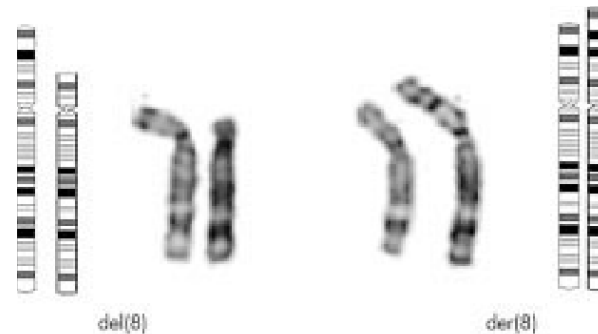
Property of  
Lamberto Camurri Ph.D.

DNA  
Fetale nel  
Plasma  
Materno

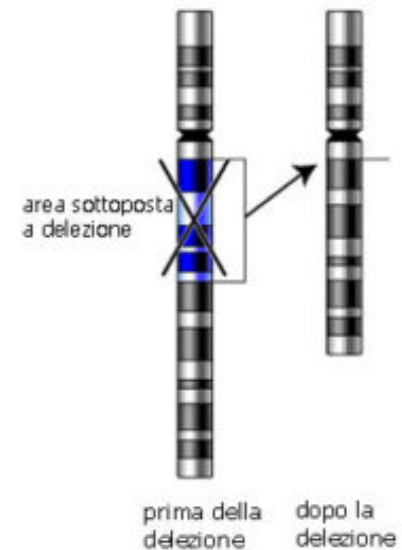


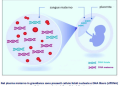
Anomalie Cromosomiche, trisomie e anomalie struttura  
Cariotipo molecolare. 46 cromosomi

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

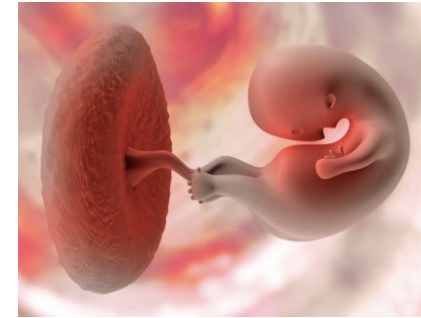
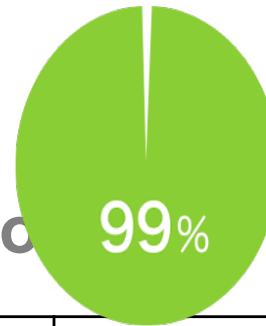


LABORATORIO BIOGEN





# NIPT DNA fetale in sangue materno

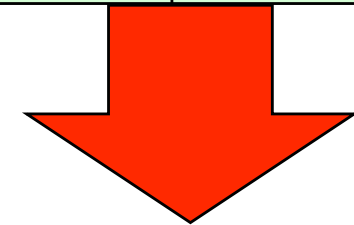


Casi di Anomalie Cromosomiche	% prevalenza EU	Validazione clinica	cffDNA Sangue materno		cffDNA Sangue materno		DNA fetale placenta	Liquido Amniotico
			SP	PPV	SN	NPV	Accuratezza%	Accuratezza%
<b>T21 T18 T13</b>	<b>3,1</b>	<b>casi &gt; 72000</b>	99,9 99,9 99,9	99 99 82	99,5 99,9 99,9	99 99 99	99,96 99,87 99,92	100

<b>XXX,XXY,XYX,X</b>	<b>0,6</b>	<b>casi &gt; 72000</b>	99,8- 99,9	<b>99-80</b>	100 -98	100
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SP	PPV	SE	NPV
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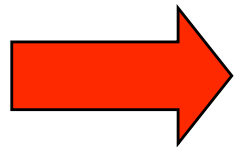
<b>Anomalie &gt; 10Mb</b>	<b>0,4</b>	<b>casi &gt; 72000</b>	99,9	<b>55,6</b>	99- 100	100
---------------------------	------------	------------------------	------	-------------	------------	-----



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

## Ricerca anomalie fetali con cffDNA: performances

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



**Falsi positivi >  
anomalia che non c'è**

**Falsi negativi >  
tutto bene ma non è vero**

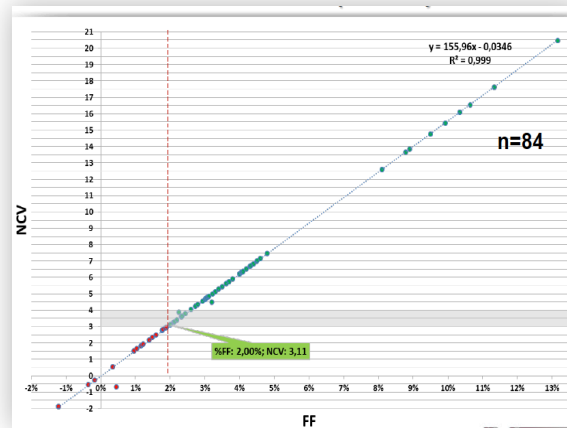
# Ricerca anomalie cromosomi con cffDNA



## Trisomie & Cario

### analisi diretta del cariotipo molecolare fetale

#### SAFER SCORE



#### RISULTATO NEGATIVO.

L'analisi del DNA fetale libero (cffDNA) eseguita sul campione biologico in esame **NON HA RILEVATO** aneuploidie o alterazioni cromosomiche strutturali, nei limiti di risoluzione della metodica.

Sesso fetale: **Maschio.**

Frazione fetale: **10%**

Rischio di risultati Falsi Negativi: **<0.1%**

L'analisi del DNA fetale libero (cffDNA) eseguita sul campione biologico in esame **HA RILEVATO una SOSPETTA DUPLICAZIONE del braccio corto del cromosoma 12.**

Non sono state rilevate aneuploidie o alterazioni cromosomiche strutturali a carico di altri cromosomi, nei limiti di risoluzione della metodica.

Sesso fetale: **Femmina.**

Frazione fetale: **11%**

Valore Predittivo Positivo (PPV): **61.54%**



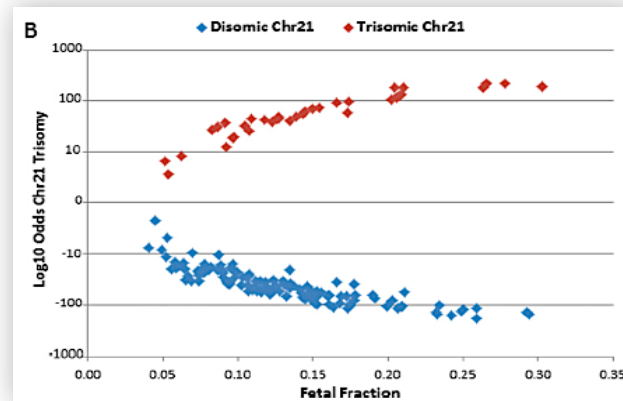
# Ricerca anomalie cromosomi con cffDNA

Harmony™  
PRENATAL TEST

VANADIS®

## Trisomie

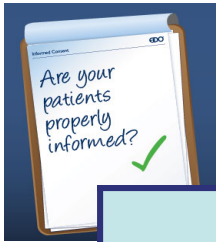
elaborazione del rischio con età, peso e altezza  
indicazione di rischio



Risultati del test		Percentuale fetale di cfDNA: 20%	
CROMOSOMA	RISULTATO	PROBABILITÀ	RACCOMANDAZIONI
Trisomia 21 (T21)	Rischio Basso	Inferiore a 1/10.000 (0,01%)	Rivedere i risultati con la paziente
Trisomia 18 (T18)	Rischio Basso	Inferiore a 1/10.000 (0,01%)	Rivedere i risultati con la paziente
Trisomia 13 (T13)	Rischio Basso	Inferiore a 1/10.000 (0,01%)	Rivedere i risultati con la paziente
Analisi Y	Feto Femminile	Superiore a 99/100 (99%)	Rivedere i risultati con la paziente

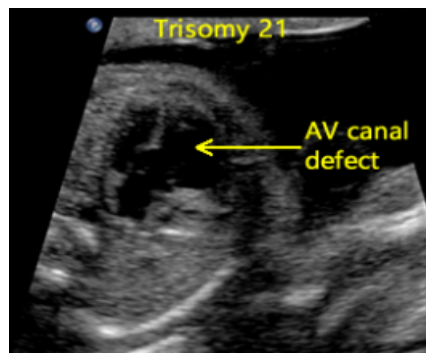
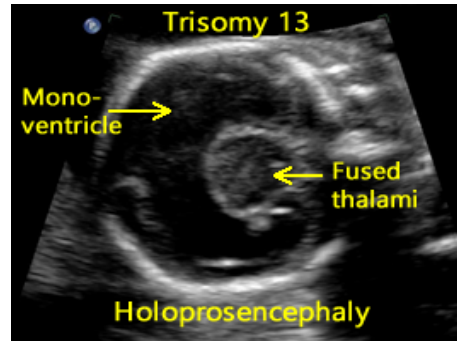
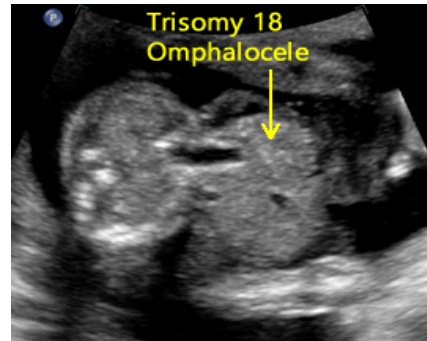
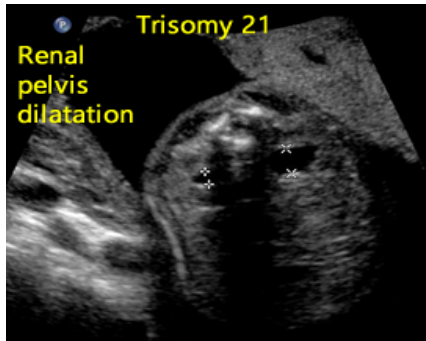
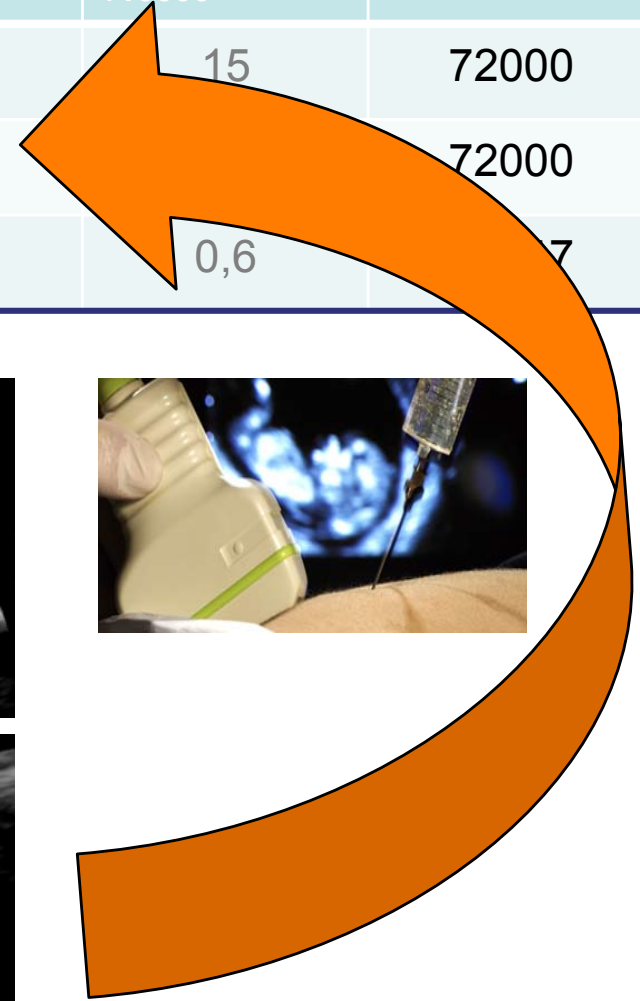
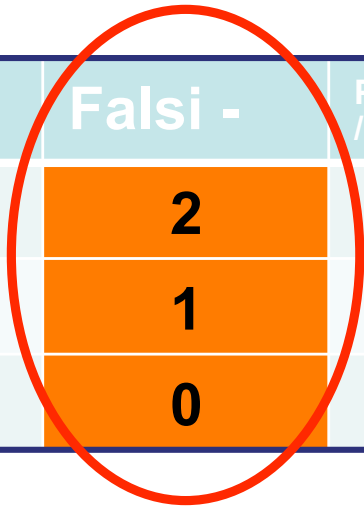
Risultati del test		Percentuale fetale di cfDNA: 9,7%	
CROMOSOMA	RISULTATO	PROBABILITÀ	RACCOMANDAZIONI
Trisomia 21 (T21)	Rischio Alto	23/100 (23%)	Consulenza genetica e test supplementari
Trisomia 18 (T18)	Rischio Basso	Inferiore a 1/10.000 (0,01%)	Rivedere i risultati con la paziente
Trisomia 13 (T13)	Rischio Basso	Inferiore a 1/10.000 (0,01%)	Rivedere i risultati con la paziente

Probability scale for chromosome 21: <math>< 1/10.000 (0,01\%)</math>, <math>1/1.000 (0,1\%)</math>, <math>1/100 (1\%)</math>, <math>10/100 (10\%)</math>, <math>50/100 (50\%)</math>, <math>90/100 (90\%)</math>, <math>> 99/100 (99\%)</math>. A marker for chromosome 21 is positioned at approximately 23% probability.



# Ricerca trisomie fetali con cffDNA: performances

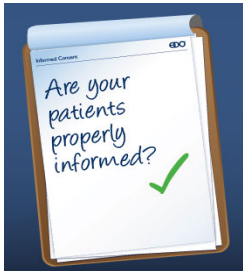
	Falsi +	Veri +	Falsi -	Prev-coorte /10000	Prev-popolaz /10000	Casi coorte
<b>T21</b>			<b>2</b>	120	15	72000
<b>T18</b>			<b>1</b>	30		72000
<b>T13</b>			<b>0</b>	13	0,6	7



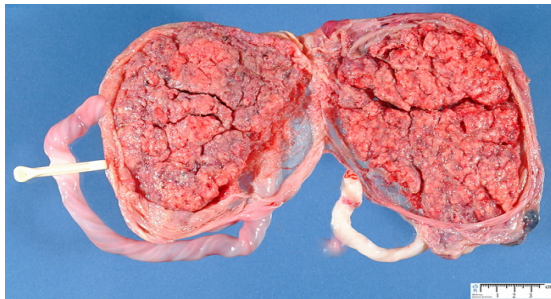
# Fetal Aneuploidy Detection with cffDNA

## 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

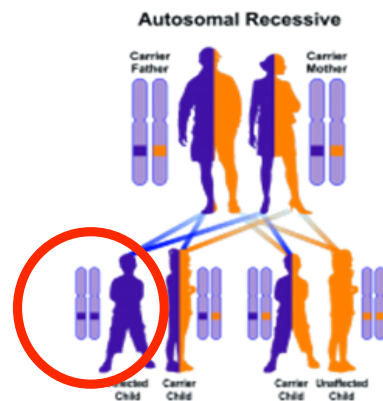


# Test Prenatale Non Invasivo. Malattie genetiche: frequenti, gravi.

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: prollasso della mitrale, lassità articolare, piede piatto
- Disfunzioni ipotalamiche?

## Test Prenatale Non Invasivo. Malattie genetiche: 300 geni

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



### La genomica di nuova generazione

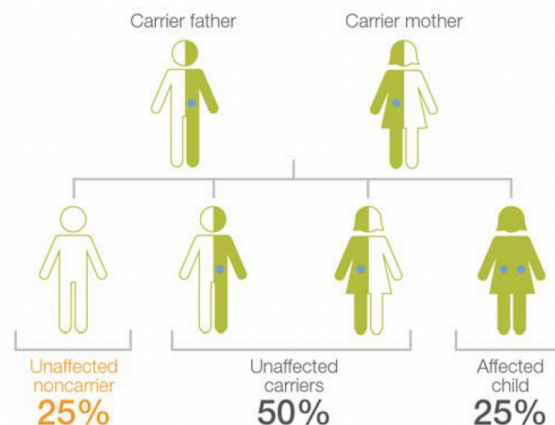
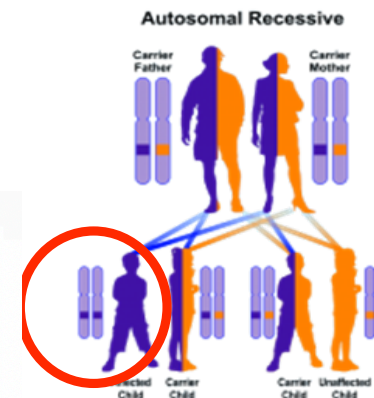
Negli ultimi anni, gli straordinari progressi conseguiti nel settore della genomica e delle biotecnologie hanno posto le basi per leggere e comprendere le informazioni contenute nel nostro DNA, il **genoma**. In particolare **le nuove tecnologie di sequenziamento, Next Generation Sequencing (NGS)**, 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.

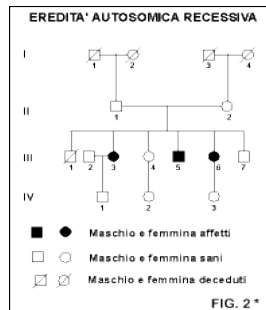
I portatori di malattie genetiche sono tipicamente individui sani, completamente privi di sintomi ed inconsapevoli di essere a rischio di trasmettere tale "errore" del DNA ai figli.

### Il test GeneScreen® Easy

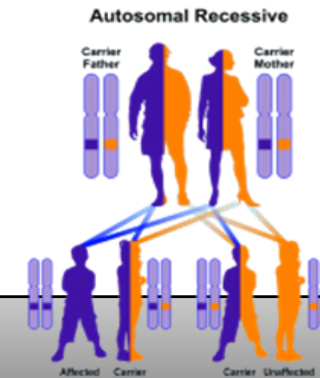
**GeneScreen® Easy** è un test diagnostico, sviluppato da GENOMA Group, che permette di eseguire un'analisi multipla di **oltre 600 malattie genetiche ereditarie**, tra cui quelle più frequenti nella popolazione italiana, come la Fibrosi Cistica, l'Anemia Falciforme, la Talassemia, la Sordità Ereditaria.



Test  
Prenatale  
Non  
Invasivo



PluriGenTEST™



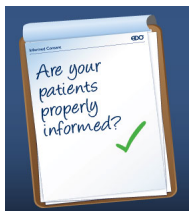
Ricerca portatori malattie mendeliane

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

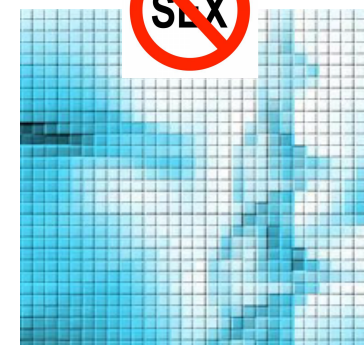
Probabilità feto affetto malattie mendeliane recessive

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

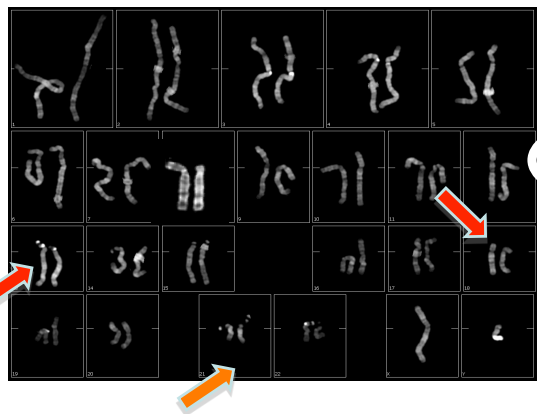
Mutazioni: Neg 1(37), Neg3 (139-152), Neg4 (intero gene)

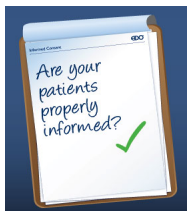


# Prenatal screening



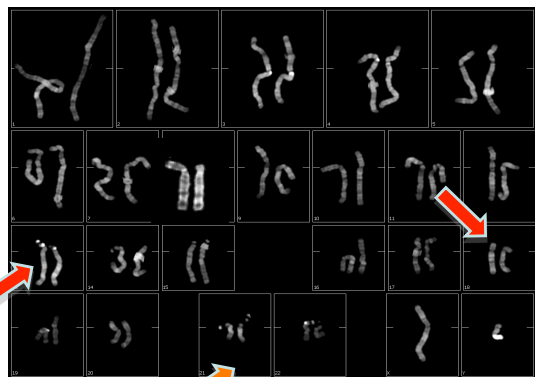
Anomalia	Anomalie ‰ nati	Specificità Sensibilità	Costo
Genetiche ereditarie	1,5	100 75-99	
Trisomie 21,18,13	3,1	99,7-99,9 98,4-99,5	650*
Cariotipo molecolare	4,1	99,9 (ppv 61) 99	
Microdelezioni 9 sindromi		- 50-95	
Altre malattie genetiche ereditarie SMA, sordità, talassemia	0,5	100 92-99	
Malattie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





# Prenatal screening

Fibrosi Cistica 90%  
 Ritardo mentale FRAXA 99%  
 Atrofia Muscolare Spinale 93%



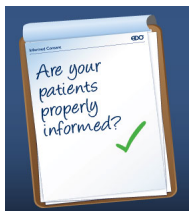
Anomalia	Anomalie ‰ nati	Specificità Sensibilità	Costo
Genetiche ereditarie	1,5	100 75-99	590
Trisomie 21,18,13	3,1	99,7-99,9 98,4-99,5	650
Cariotipo molecolare	4,1	99,9 (ppv 61) 99	
Microdelezioni 9 sindromi		- 50-95	
Altre malattie genetiche ereditarie sordità, talassemia	0,5	100 92-99	
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	

PluriGenTEST™



PluriGenTEST™





# Prenatal screening

Fibrosi Cistica 90%  
 Ritardo mentale FRAXA 99%  
 Atrofia Muscolare Spinale 93%



Anomalia	Anomalia % nati	Specificità Sensibilità	Costo
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
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	

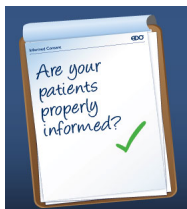
PluriGenTEST™

PRENATAL EASY



PluriGenTEST™

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 Lamberto Camurri Ph.D.



# Prenatal screening

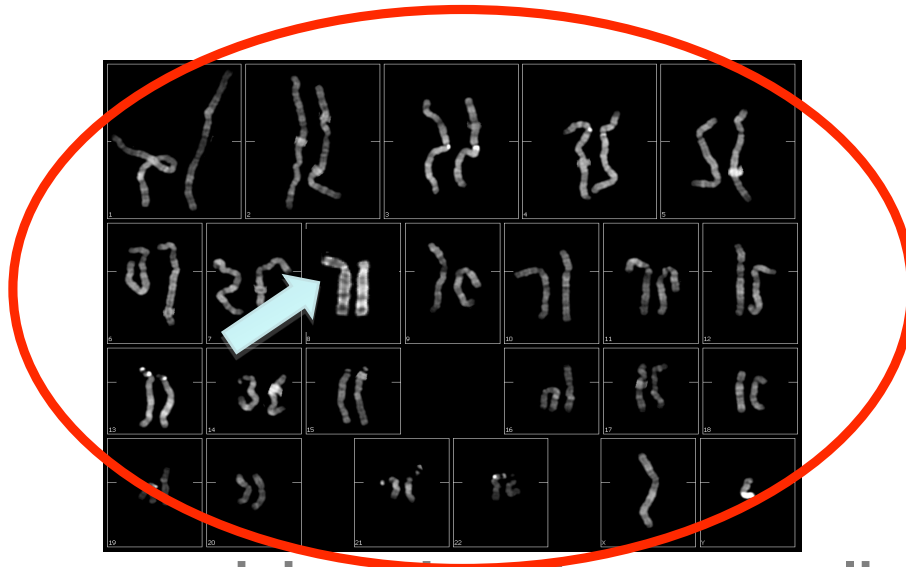
Anomalia	Anomalia ‰ nati	Specificità Sensibilità	Costo
<b>Genetiche ereditarie</b>	<b>1,5</b>	<b>100</b> 75-99	
<b>Trisomie 21,18,13</b>	<b>3,1</b>	<b>99,7-99,9</b> 98,4-99,5	
<b>Cariotipo molecolare comprese T21,18,13</b>	<b>4,1</b>	<b>99,9 (ppv 61)</b> 99	<b>895</b>
<b>Microdelezioni 9 sindromi</b>		<b>-</b> 50-95	
<b>Altre malattie genetiche ereditarie sordità, talassemia, 300 geni</b>	<b>2,5</b>	<b>100</b> 92-99	
<b>Malattie genetiche non ereditarie</b>	<b>&lt;0,01-0,4</b>	<b>&gt;95</b> >70	



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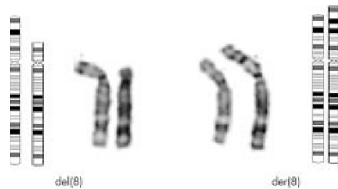
**DNA  
Fetale nel  
Plasma  
Materno**

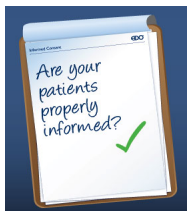


**Anomalie Cromosomiche, trisomie e anomalie struttura  
46 cromosomi**

Sopravvivenza. 1anno, 10 anni

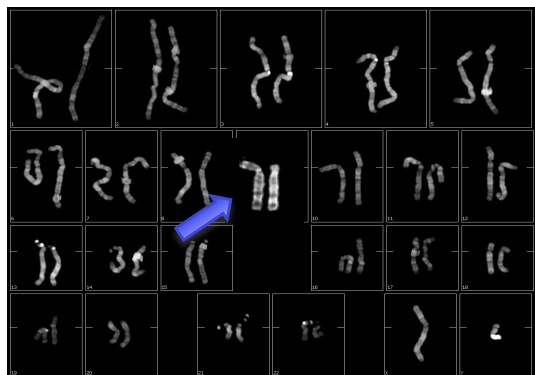
<b>DELEZIONI</b>	<b>0,30‰ (1/3000)</b>	1° 90%	10° >50%
<b>DUPLICAZIONI</b>	<b>0,15 ‰ (1/6000)</b>	1° 90%	10° >50%





# Prenatal screening

Fibrosi Cistica 90%  
 Ritardo mentale FRAXA 99%  
 Atrofia Muscolare Spinale 93%



Sordità Neurosensoriale 90%

Anomalia	Anomalia ‰ nati	Specificità Sensibilità	Costo
Genetiche ereditarie	1,5	100 75-99	590
Trisomie 21,18,13	3,1	99,7-99,9 98,4-99,5	
Cariotipo molecolare	4,1	99,9 (ppv 61) 99	895
Microdelezioni 9 sindromi		- 50-95	200
Altre malattie genetiche ereditarie Genescreen 300	2,5	100 92-99	740
Malattie genetiche Genesafe denovo	<0,01-0,4	>95 >70	740

PluriGenTEST™

PRENATAL  
EASY

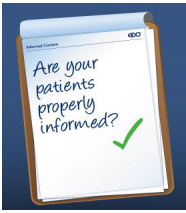
plus

genescreen®

GeneSafe™

PluriGenTEST™

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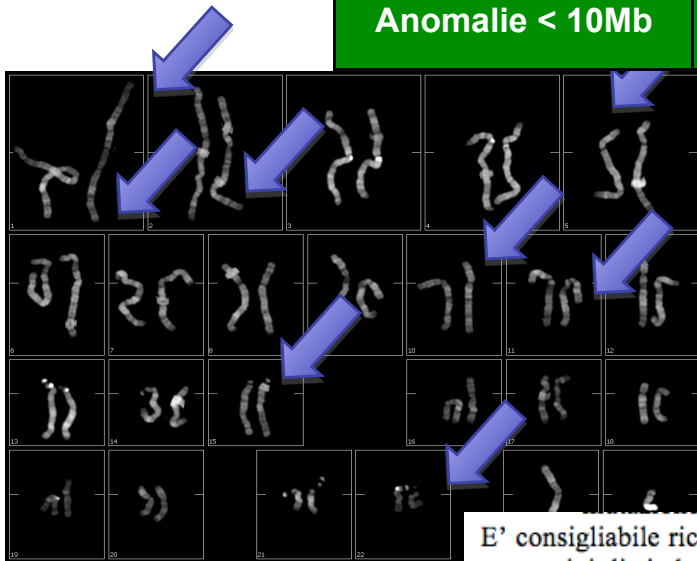


# Prenatal screening

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.

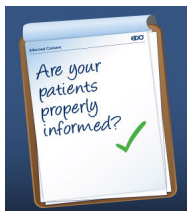


Casi di Anomalie Cromosomiche	Popolazione EU	% prevalence	% Anomalie Cromosomiche
<b>Totale</b>	<b>10323</b>	<b>4,4</b>	
<b>Anomalie cromosomiche strutt.</b>	<b>1737</b>	<b>0,7</b>	<b>17 (40&lt;10)</b>
<b>Anomalie &lt; 10Mb</b>			<b>3</b>



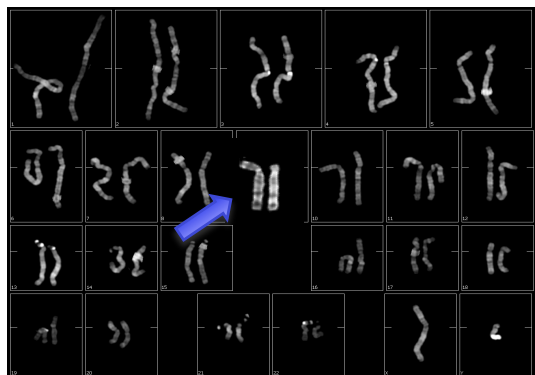
Sindrome da microdelezione	Regione cromosomica	Prevalenza (alla nascita)
Sindrome di DiGeorge	delezione 22q11.2	1/2.000 - 1/4.000
Sindrome Cri-du-chat	delezione 5p15.3	1/15.000 - 1/50.000
Sindrome di Prader-Willi	delezione 15q11.2	1/25.000
Sindrome Angelman	delezione 15q11.2	1/10.000 - 1/20.000
Sindrome 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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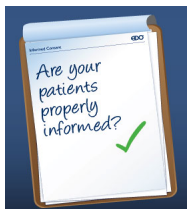


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# Prenatal screening

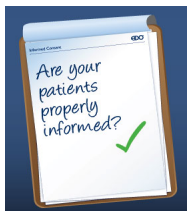
Malattie genetiche	Anomalie %	Specificità Sensibilità BIOLOGICA	Specificità Sensibilità TECNICA	Anomalie % tot
Malattie genetiche non ereditarie	<0,01-0,4	>95 >70	>95 >70	



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-Giadion
SINDROME DI NOONAN	
BRAF	Sindrome Cardio facio cutanea (CFS) tipo 1 1/2500 Se 50-75%
CBL	Sindrome di Noonan-simile con o senza leucemia mielomonocitica giovanile
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 Acondroplasia Sindrome CATSHL Sindromedi Crouzon con acanthosis nigricans
FGFR3	Ipocondroplasia Sindrome di Muenke Displasia tanatofora, tipo I Displasia tanatofora, tipo II Sindrome di Ehlers-Danlos, classica Sindrome di Ehlers-Danlos, tipo VIIA
COL1A1	Osteogenesi imperfetta, tipo I Osteogenesi imperfetta, tipo II 1/20000 Se 95% Osteogenesi imperfetta, tipo III Osteogenesi imperfetta, tipo IV Sindrome di Ehlers-Danlos, forma cardiaco-valvolare Sindrome di Ehlers-Danlos, tipo VIIB
COL1A2	Osteogenesi imperfetta, tipo II Osteogenesi imperfetta, tipo III Osteogenesi imperfetta, tipo IV
CRANIOSINOSTOSI	
FGFR2	Sindrome di Antley-Bixler senza anomalie genitali o disordini della steroidognesi Sindrome di Apert 1/2500 Se 20% 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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# Prenatal screening

Malattie genetiche	Anomalia %	S S B
	1-0,4	

## Short stature

- Up to 83% of patients have short stature<sup>1</sup>

## Characteristic facial features<sup>2</sup>

- Broad, high forehead
- Hypertelorism
- Low-set, posteriorly rotated ears with a thick helix
- High-arched palate
- Micrognathia
- High-arched eyebrows<sup>1</sup>
- Short neck with excess nuchal skin
- Epicanthal folds
- Downward-slanting palpebral fissures
- Low posterior hairline

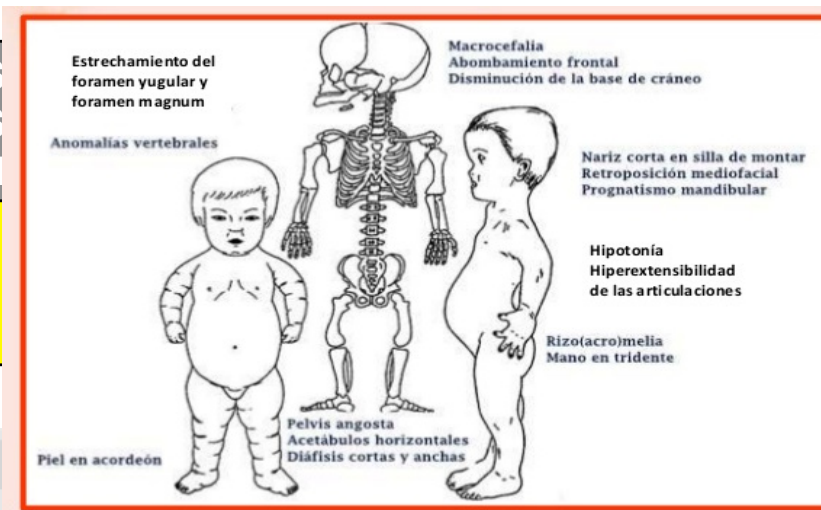


## Congenital heart defects<sup>3</sup>

- Pulmonary valve stenosis
- Hypertrophic obstructive cardiomyopathy
- Atrial and ventricular septal defects
- Persistent ductus arteriosus

## Other clinical manifestations<sup>3-5</sup>

- Pectus carinatum, pectus excavatum
- Scoliosis
- Cryptorchidism
- Lymphatic abnormalities
- Coagulopathy
- Cognitive/learning disabilities
- Ophthalmological issues
- Arnold-Chiari malformation
- Seizures



## GENE

COL2A1

- Acondroplasia
- Síndrome CATSHL
- Síndromedi Crouzon con acanthosis nigricans
- Ipocondroplasia
- Síndrome di Muenke

FGFR3

COL1A1



Síndrome di Pfeiffer, tipo 2

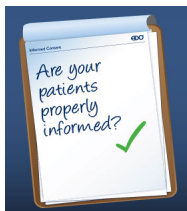
Síndrome di Pfeiffer, tipo 3

FGFR2



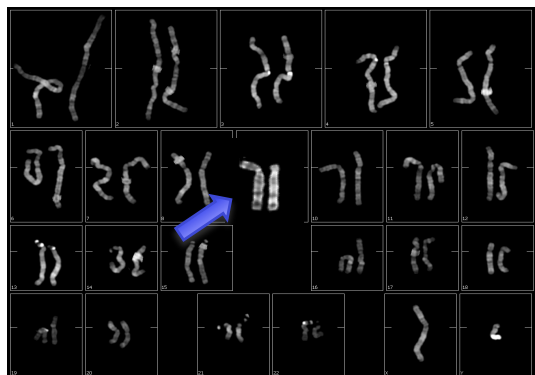
SINDROME DI NOONAN	
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<b>CBL</b>	Síndrome di Noonan-simile con o senza leucemia mielomonocítica giovanile
<b>KRAS</b>	Síndrome di Noonan /cancers
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<b>MAP2K2</b>	Síndrome Cardio facio cutanea (CFS) tipo 4
<b>NRAS</b>	Síndrome di Noonan 6/cancers
<b>PTPN11</b>	Síndrome Noonan 1/ Síndrome di LEOPARD/cancers
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<b>RAF1</b>	Síndrome di Noonan 5/Síndrome di LEOPARD 2
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<b>SOS1</b>	Síndrome di Noonan 4





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

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

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

Pag. 7 di 26

Eurofins Genoma Group S.r.l a socio unico

Sede legale  
00138 Roma - Via di Castel Giubileo, 11  
C.F. e P.Iva: 05402921000  
REA: 883.955  
Iscr. Reg. Impr. 368761/1987

Laboratori e Studi Medici Roma  
00138 Roma - Via di Castel Giubileo, 11  
Tel. +39 06 881 1270 - Fax +39 06 6449 2025  
Web: www.laboratoriongenoma.eu  
E-mail: info@laboratoriongenoma.eu

Laboratori e Studi Medici Milano  
20161 Milano - Affori Centre, Via Enrico Cialdini, 16  
Tel. : + 39 02 3929 7626 - Fax: + 39 02 3929 76261  
Web: www.genomamilano.it  
E-mail: info@genomamilano.it

CHIAMATA GRATUITA  
NUMEROVERDE  
800-501651

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

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Sede legale  
00138 Roma - Via di Castel Giubileo, 11  
C.F. e P.Iva: 05402921000  
REA: 883.955  
Iscr. Reg. Impr. 368761/1987

Laboratori e Studi Medici Roma  
00138 Roma - Via di Castel Giubileo, 11  
Tel. +39 06 881 1270 - Fax +39 06 6449 2025  
Web: www.laboratoriongenoma.eu  
E-mail: info@laboratoriongenoma.eu

Laboratori e Studi Medici Milano  
20161 Milano - Affori Centre, Via Enrico Cialdini, 16  
Tel. : + 39 02 3929 7626 - Fax: + 39 02 3929 76261  
Web: www.genomamilano.it  
E-mail: info@genomamilano.it

CHIAMATA GRATUITA  
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800-501651

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

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

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

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

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

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

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

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

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

LYST	<a href="#">606897</a>	Chediak-Higashi syndrome	<a href="#">214500</a>	AR
MAN2B1	<a href="#">609458</a>	Mannosidosis, alpha-, types I and II	<a href="#">248500</a>	AR
MCCC1	<a href="#">609010</a>	3-Methylcrotonyl-CoA carboxylase 1 deficiency	<a href="#">210200</a>	AR
MCCC2	<a href="#">609014</a>	3-Methylcrotonyl-CoA carboxylase 2 deficiency	<a href="#">210210</a>	AR
MCOLN1	<a href="#">605248</a>	Mucopolipidosis IV	<a href="#">252650</a>	AR
MECP2	<a href="#">300005</a>	Encephalopathy, neonatal severe	<a href="#">300673</a>	XLR
		Mental retardation, X-linked syndromic, Lubs type	<a href="#">300260</a>	XLR
		Mental retardation, X-linked, syndromic 13	<a href="#">300055</a>	XLR
		Rettsyndrome	<a href="#">312750</a>	XLD
		Rettsyndrome, atypical	<a href="#">312750</a>	XLD
		Rettsyndrome, preserved speech variant	<a href="#">312750</a>	XLD
		Autism susceptibility, X-linked 3	<a href="#">300496</a>	XL
MED17	<a href="#">603810</a>	Microcephaly, postnatal progressive, with seizures and brain atrophy	<a href="#">613668</a>	AR
MEFV	<a href="#">608107</a>	Familial Mediterranean fever, AD	<a href="#">134610</a>	AD
		Familial Mediterranean fever, AR	<a href="#">249100</a>	AR
MFSB8	<a href="#">611124</a>	Ceroid lipofuscinosis, neuronal, 7	<a href="#">610951</a>	AR
		Macular dystrophy with central cone involvement	<a href="#">616170</a>	AR
MKS1	<a href="#">609883</a>	Bardet-Biedl syndrome 13	<a href="#">615990</a>	AR
		Joubert syndrome 28	<a href="#">617121</a>	AR
		Meckel syndrome 1	<a href="#">249000</a>	AR
MLC1	<a href="#">605908</a>	Megalencephalic leukoencephalopathy with subcortical cysts	<a href="#">604004</a>	AR
MMAA	<a href="#">607481</a>	Methylmalonic aciduria, vitamin B12-responsive	<a href="#">251100</a>	AR
MMAB	<a href="#">607568</a>	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cb1B complementation type	<a href="#">251110</a>	AR
MMACHC	<a href="#">609831</a>	Methylmalonic aciduria and homocystinuria, cb1C type	<a href="#">277400</a>	AR
MMADHC	<a href="#">611935</a>	Homocystinuria, cb1D type, variant 1	<a href="#">277410</a>	AR
MPI	<a href="#">154550</a>	Congenital disorder of glycosylation, type Ib	<a href="#">602579</a>	AR
MPL	<a href="#">159530</a>	Myelofibrosis with myeloid metaplasia, somatic	<a href="#">254450</a>	
		Thrombocythemia 2	<a href="#">601977</a>	SMu, AD



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

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

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

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

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

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Eurofins Genoma Group S.r.l a socio unico

Sede legale  
00138 Roma - Via di Castel Giubileo, 11  
C.F. e P.Iva: 05402921000  
REA: 883.955  
Iscr. Reg. Impr. 368761/1987

Laboratori e Studi Medici Roma  
00138 Roma - Via di Castel Giubileo, 11  
Tel. +39 06 881 1270 - Fax +39 06 6448 2025  
Web: www.laboratoriongenoma.eu  
E-mail: info@laboratoriongenoma.eu

Laboratori e Studi Medici Milano  
20161 Milano - Affori Centre, Via Enrico Cialdini, 16  
Tel. : + 39 02 3929 7626 - Fax: + 39 02 3929 76261  
Web: www.genomamilano.it  
E-mail: info@genomamilano.it

CHIAMATA GRATUITA  
NUMEROVERDE  
800-501651

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

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Sede legale  
00138 Roma - Via di Castel Giubileo, 11  
C.F. e P.Iva: 05402921000  
REA: 883.955  
Iscr. Reg. Impr. 368761/1987

Laboratori e Studi Medici Roma  
00138 Roma - Via di Castel Giubileo, 11  
Tel. +39 06 881 1270 - Fax +39 06 6448 2025  
Web: www.laboratoriongenoma.eu  
E-mail: info@laboratoriongenoma.eu

Laboratori e Studi Medici Milano  
20161 Milano - Affori Centre, Via Enrico Cialdini, 16  
Tel. : + 39 02 3929 7626 - Fax: + 39 02 3929 76261  
Web: www.genomamilano.it  
E-mail: info@genomamilano.it

CHIAMATA GRATUITA  
NUMEROVERDE  
800-501651

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

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