

# PRENATAL REQUISITION FORM



## Page 1 of 2

Please complete all information below.  
Failure to do so may delay sample processing.  
We accept samples Monday through Saturday.

2820 North Astor Street, Spokane, WA 99207  
Phone: 509.474.6840 / Fax: 509.474.6839  
TOLL FREE 1.877.SigChip (744.2447)  
www.signaturegenomics.com / info@signaturegenomics.com

### INFORMED CONSENT REQUIRED

NOTE TO HEALTH CARE PROVIDERS: Signature Genomics recognizes that it is important for physicians to discuss pertinent information about genetic testing with their patients. In accordance with Washington State Law RCW 7.70.050 and WAC 388-531-0050, the physician referring the patient for laboratory testing is responsible for informing the patient of the risks and benefits of the laboratory testing and obtaining the patient's informed consent. As a courtesy to our ordering health care providers and their patients, we offer educational literature including *Points to Consider*, *Frequently Asked Questions for Physicians*, *Frequently Asked Questions for Prenatal Testing* and *Informed Consent for Molecular Cytogenetic Testing* to assist in discussions of the benefits and limitations of array CGH testing. To obtain copies of these documents and other helpful resources, please visit our website at [www.signaturegenomics.com](http://www.signaturegenomics.com) or contact us directly at 1.877.SigChip (1.877.744.2447).

By signing below, I attest that I have obtained informed consent for the test(s) that I have ordered below through Signature Genomics from this patient and/or guardian(s).

PHYSICIAN SIGNATURE: \_\_\_\_\_ *Required for samples originating in New York*

### PATIENT INFORMATION

DOB \_\_\_\_ / \_\_\_\_ / \_\_\_\_ (mm dd yyyy)     Male     Female  
Last Name \_\_\_\_\_  
First Name \_\_\_\_\_ M.I. \_\_\_\_\_  
Address \_\_\_\_\_  
City, State, ZIP \_\_\_\_\_  
Home (\_\_\_\_) \_\_\_\_\_ Work (\_\_\_\_) \_\_\_\_\_  
Specimen # / ID # \_\_\_\_\_

### PREGNANCY INFORMATION

I. Is this an on-going pregnancy?     Yes     No  
II. Fetal Sex (required for accurate analysis)     Male     Female     Unknown  
III. Karyotype     Normal     Abnormal     Pending     Not Performed  
Details \_\_\_\_\_ (attach report)  
IV. Weeks Gestation \_\_\_\_\_     by LMP \_\_\_\_\_     by Ultrasound on \_\_\_\_ / \_\_\_\_ / \_\_\_\_

### INDICATION FOR STUDY (IFS)

I. Check all that apply and attach or provide additional details below  
 Abnormal maternal serum screen     Family history  
 Abnormal ultrasound     Parental concern  
 Advanced maternal age     Other

Provide additional details \_\_\_\_\_

II. ICD-9 code (required for insurance billing) \_\_\_\_\_

### PRENATAL SAMPLE TEST REQUESTED

Signature PrenatalChip® (Targeted)  
 SignatureChipWG™ (Whole Genome)  
 SignatureChipOS™ (Oligo Solution)  
 Single/Dual FISH: Contact Signature Genomics to discuss case.  
 Karyotype (Note: If karyotype is not requested it should be performed elsewhere for all prenatal specimens)

#### Amniotic fluid only (choose one):

AFP only     AChE only     AFP & AChE     AFP w/reflex to AChE

#### Maternal Cell Contamination Studies

Signature Genomics strongly recommends that MCC testing be performed on each prenatal sample that is received. If MCC is pending or has already been performed at a different laboratory, please check this box  and forward test results. Note that performing MCC studies on a different sample from the same source is not enough to rule out MCC. There is an additional charge for MCC studies. Requires 3-5cc maternal blood.

Proceed with MCC     Decline MCC

### PARENTS' SAMPLES

Check one. Please note that charges may apply.

Fetal sample to accompany or follow.  
 Fetal analysis complete. Perform follow-up analysis on parental sample(s). If Signature Genomics is to bill a new insurance, please fill out page 2 of this form.

Sample(s) included in this shipment:

**Mother**  
Name \_\_\_\_\_ DOB \_\_\_\_ / \_\_\_\_ / \_\_\_\_  
Sample Type     Blood     DNA     other \_\_\_\_\_  
 **Father**  
Name \_\_\_\_\_ DOB \_\_\_\_ / \_\_\_\_ / \_\_\_\_  
Sample Type     Blood     DNA     other \_\_\_\_\_

### REFERRING PHYSICIAN

Name \_\_\_\_\_  
NPI# \_\_\_\_\_ Specialty \_\_\_\_\_  
Address \_\_\_\_\_  
City, State, ZIP \_\_\_\_\_  
Hospital/Institution \_\_\_\_\_  
Tel (\_\_\_\_) \_\_\_\_\_ FAX (\_\_\_\_) \_\_\_\_\_  
Counselor/Contact \_\_\_\_\_  
Tel (\_\_\_\_) \_\_\_\_\_ FAX (\_\_\_\_) \_\_\_\_\_  
Address \_\_\_\_\_  
City, State, ZIP \_\_\_\_\_  
E-mail \_\_\_\_\_  
**Additional Reports To:**  
Name \_\_\_\_\_  
Hospital/Institution \_\_\_\_\_  
Address \_\_\_\_\_  
City, State, ZIP \_\_\_\_\_  
Tel (\_\_\_\_) \_\_\_\_\_ FAX (\_\_\_\_) \_\_\_\_\_

### SAMPLE INFORMATION

Indicate the reference lab sending sample to Signature Genomics:

Date Sample Obtained \_\_\_\_ / \_\_\_\_ / \_\_\_\_ (mm dd yyyy)

#### CVS

Cultured cells

#### AMNIOCENTESIS

Direct fluid     Cultured cells

#### PRODUCTS OF CONCEPTION

Specify tissue of origin \_\_\_\_\_  
 Cultured cells     Direct specimen

#### BLOOD

Fetal, specify source:     PUBS     Cord blood – post delivery

**DNA** Please read *DNA Acceptance Policy* at [www.signaturegenomics.com](http://www.signaturegenomics.com)

Specify DNA source:    direct    cultured

CVS	<input type="checkbox"/>	<input type="checkbox"/>
Amnio	<input type="checkbox"/>	<input type="checkbox"/>
Tissue	<input type="checkbox"/>	<input type="checkbox"/>
Blood (PUBS)	<input type="checkbox"/>	<input type="checkbox"/>
Blood (cord)	<input type="checkbox"/>	<input type="checkbox"/>

#### OTHER

Please specify \_\_\_\_\_

#### Specimen Requirements

**Cultured cells:** 1x T-75 flask or 3x T-25 flasks, 70% confluent

**Direct amniotic fluid:** 15-20cc

**Tissue/POC:** 15-20 mg tissue in sterile media or saline

**Fetal blood:** Minimum 1cc in NaHep

**Parental Blood:** 2 tubes, 1x 3-5cc EDTA and 1x 3-5cc NaHep

*Additional costs may be incurred if specimen quality or quantity is suboptimal.*

### USE OF SPECIMENS

Signature Genomics (SG) retains patient samples indefinitely for validation, educational purposes and/or research, maintaining the confidentiality of each sample. Patients may decline the use of submitted sample(s) for research; refusal does not impact diagnostic testing or reporting of results. Patients may withdraw consent for use of sample(s) at any time by contacting the SG Chief Medical Officer at (509) 474-6840. SG will not pay royalties to patients if a commercial product is developed during research using their samples. I do not wish to allow my (or my fetus's) sample to be used for test validation or education. Therefore, I am checking this box  to indicate that the sample should be used for the test specified above and will be destroyed after 60 days.

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## Page 2 of 2

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### PATIENT INFORMATION

DOB \_\_\_\_/\_\_\_\_/\_\_\_\_ (mm dd yyyy)  
Name \_\_\_\_\_  
Last First

### INSTITUTIONAL BILLING

P.O. \_\_\_\_\_  
Client ID \_\_\_\_\_  
Institution Name \_\_\_\_\_  
Financial Contact \_\_\_\_\_  
Address \_\_\_\_\_  
City, State, ZIP \_\_\_\_\_  
TEL (\_\_\_\_) \_\_\_\_\_ FAX (\_\_\_\_) \_\_\_\_\_

### SELF-PAY

- Check or Money Order  Credit Card  
 Send receipt to patient address on Page 1

Payment will not be processed before sample arrives. This is to ensure that the correct amount is charged to the credit card.

**Check one:**  AMEX  Visa  MC  Discover

Acct # \_\_\_\_\_ Exp Date \_\_\_\_/\_\_\_\_  
(mm yyyy)

Zip Code \_\_\_\_\_ 3 or 4 Digit Security Code \_\_\_\_\_

Print Name as it appears on card \_\_\_\_\_

Signature \_\_\_\_\_ Date \_\_\_\_/\_\_\_\_/\_\_\_\_  
(mm dd yyyy)

A new authorization and/or signature may be required when testing additional family members.

### REQUEST FOR SIGNATURE GENOMICS TO DIRECTLY BILL PATIENT'S INSURANCE

**IMPORTANT:** Patients and health care providers requesting that Signature Genomics bill private insurance must complete this portion of the requisition form prior to or at the time of sample submission. **Failure to do so will delay testing and results.**

I have obtained prior authorization.

CPT codes and units authorized: \_\_\_\_\_

Prior-Authorization #: \_\_\_\_\_

As a courtesy service, Signature Genomics will obtain benefit information from the patient's insurance. If a benefit investigation and/or prior authorization is requested prior to sample submission, complete and fax both page 1 and page 2 of this requisition form to 509.474.6839. This information may also be submitted with the sample. Please include a Letter of Medical Necessity and clinic notes as this information may be required by the insurance company for prior authorization. For benefit investigation of parents' sample(s), please complete an additional billing page if one parent's insurance differs from the proband's insurance.

Whom should our billing specialist contact in order to communicate benefit information?  
CHECK ALL THAT APPLY (REQUIRED):  Patient  Physician  Genetic Counselor/Contact

### INSURANCE INFORMATION **Include enlarged copy of insurance card/s (front and back)**

If your institution's intake form addresses all items below, you may attach it. Otherwise, please fill in each item completely.

Is this a Medicaid Policy?  Yes  No

Insurance Company Name \_\_\_\_\_

Policyholder's Name \_\_\_\_\_  
Last First M.I.

Insurance Company Phone \_\_\_\_\_

SSN \_\_\_\_\_

Plan Name \_\_\_\_\_

Policyholder's Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_  
mm dd yyyy

ID# \_\_\_\_\_

Relationship to Patient: \_\_\_\_\_

Group# \_\_\_\_\_

If HMO, provide PCP Name \_\_\_\_\_

### AUTHORIZATION TO ASSIGN BENEFITS AND ACCEPT FINANCIAL RESPONSIBILITY FOR MY ACCOUNT

I assign and authorize insurance payments to Signature Genomics. I understand my insurance carrier may not approve and reimburse my medical genetic services in full due to usual and customary rate limits, benefit exclusions, coverage limits, lack of authorization, medical necessity or otherwise. I understand I am responsible for fees not paid in full, co-payments, and policy deductibles except where my liability is limited by contract or State and Federal law. A duplicate or faxed copy of this authorization is considered same as the original document.

### AUTHORIZATION TO CONTACT HEALTH INSURANCE CARRIER AND RELEASE CONFIDENTIAL MEDICAL INFORMATION

I understand Signature Genomics will contact my insurance carrier regarding coverage of genetic testing. I authorize the disclosure of insurance benefit coverage and payment information to Signature Genomics. I authorize my physician or other medical entity to release confidential medical information to Signature Genomics concerning my medical history. I authorize Signature Genomics to release confidential medical information to my health insurance carrier to facilitate reimbursement of my medical fees.

Check this box to hold testing until investigation of benefits / prior authorization is complete.  
If this box is not checked, analysis will proceed upon sample receipt.

Signature of Patient or Guardian

Printed Name of Patient or Guardian

Date

### PATIENT FINANCIAL ASSISTANCE

**FINANCIAL ASSISTANCE INFORMATION.** For more information, visit our website at [www.signaturegenomics.com](http://www.signaturegenomics.com).

Household size (e.g. number of people): \_\_\_\_\_ Household income (monthly): \_\_\_\_\_

By signing below, I certify that the household size and income information shown above is correct. Copies of tax returns, pay stubs and other information verifying income may be required before a discount is applied.

Signature of Patient or Guardian \_\_\_\_\_ Date \_\_\_\_\_

Please fax completed forms to **509.474.6839** and include in sample shipment.

# SAMPLE REQUIREMENTS AND TURN-AROUND TIMES

Phone: 509.474.6840 / Fax: 509.474.6839  
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www.signaturegenomics.com / info@signaturegenomics.com



## Signature Microarrays

SAMPLE TYPE	QUANTITY REQUIRED	TURN-AROUND TIMES*
<b>Peripheral Blood</b>	2 tubes (1 x NaHep and 1 x EDTA vacutainer) each containing 3-5 cc whole blood	5 days (WG) / 7 days (OS)
<b>Skin Biopsy</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> ) or 3mm x 3mm biopsy in sterile media or saline	CULTURED CELLS: 5 days TISSUE: <i>Please contact the Laboratory</i>
<b>DNA</b>	30µg <i>Please read DNA Acceptance Policy</i>	5 days
<b>Cultured CVS or Amniocytes</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> )	5 days
<b>Amniotic Fluid</b>	15-20 cc	<i>Please contact the Laboratory</i>
<b>Placental Tissue/ Products of Conception (POC)</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> ) or 15-20 mg tissue in sterile media or saline	CULTURED CELLS: 5 days TISSUE: <i>Please contact the Laboratory</i>

## Signature FISH

SAMPLE TYPE	QUANTITY REQUIRED	TURN-AROUND TIMES*
<b>Peripheral Blood</b>	1 NaHep vacutainer containing 3-5 cc whole blood	7-10 days
<b>Cultured CVS or Amniocytes</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> )	7-10 days
<b>Amniotic Fluid</b>	15-20 cc	<i>Please contact the Laboratory</i>
<b>Placental Tissue/ Products of Conception (POC)</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> ) or 15-20 mg tissue in sterile media or saline	CULTURED CELLS: 5 days TISSUE: <i>Please contact the Laboratory</i>

## Signature Karyotype

SAMPLE TYPE	QUANTITY REQUIRED	TURN-AROUND TIMES*
<b>Peripheral Blood</b>	1 NaHep vacutainer containing 3-5 cc whole blood	7-10 days
<b>Cultured CVS or Amniocytes</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> )	7-10 days
<b>Amniotic Fluid</b>	15-20 cc	<i>Please contact the Laboratory</i>
<b>Placental Tissue/ Products of Conception (POC)</b>	1 x T-75 flask or 3 x T-25 flasks (70% confluent <sup>‡</sup> ) or 15-20 mg tissue in sterile media or saline	CULTURED CELLS: 5 days TISSUE: <i>Please contact the Laboratory</i>

## General Specimen Handling Instructions

- Label all tubes/flasks with patient name and date of birth, and enclose completed paperwork. Include previous cytogenetic reports.
- Samples should be shipped at room temperature in a rigid, leak-proof container by overnight delivery. Delayed shipment of sample or inappropriate temperatures may result in longer processing time or sample failure. Please notify Laboratory of courier tracking number.
- Microarray turn-around times are usually 5-7 days for sufficient samples. If culturing is needed, or other services are requested, turn-around times may be longer. Samples received prior to 12:00pm (PST) are processed the same day, whereas samples received in the afternoon are processed the next working day.
- Please contact the Laboratory at 509.474.6840 for further clarification or other questions.

\* Turn-around times are based upon receipt of adequate sample for microarray analysis. Other Laboratory services, such as cell culturing, culture expansion, karyotyping, or FISH for single/dual loci, may increase turn-around times.

‡ Samples received that are less than 70% confluent will require expansion. There is an extra charge for the additional tissue culture.

# DNA ACCEPTANCE POLICY



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www.signaturegenomics.com / info@signaturegenomics.com

Signature Genomics is focused on providing clinicians and their patients with optimal results. To achieve this, Signature needs to receive DNA samples prepared under the best possible conditions.

The following are some guidelines and requirements for Signature Genomics to handle prepared patient DNA.

- We recommend using a Puregene® DNA extraction kit for whole peripheral blood samples.
- We require at least 30 micrograms of DNA preferably suspended in TE for all Signature Microarrays.
- All DNA received is re-quantified at Signature Genomics by a fluorometer prior to microarray analysis.
- All DNA received is assessed by gel electrophoresis for degradation prior to proceeding with microarray testing.
- If a sample is deemed sub-optimal for testing, the physician or counselor will be notified.
- In the event that the DNA sample is deemed to be sub-optimal and the physician still wishes to proceed with the microarray test, there will be a charge of 50% of the list price to cover Signature Genomics' processing costs if the assay fails to yield interpretable results.
- Please be advised that FISH confirmation of an abnormality seen on the microarray cannot be performed when submitting DNA only. For FISH confirmation of results please submit a harvested cytogenetic pellet along with the DNA specimen.

If you have any further questions on submitting DNA for microarray analysis please contact the laboratory.

# INSTITUTION BILLING



Phone: 509.474.6840 / Fax: 509.474.6839  
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Institution billing accounts may be established for Signature Genomics. Please complete this form and e-mail or fax it to the laboratory at 509-474-6839.

Upon receipt, an account will be established, and itemized monthly invoicing will occur.

**Institution Name:** \_\_\_\_\_

**Department or Division:** \_\_\_\_\_

**Authorized Agent:** \_\_\_\_\_

**Physicians who will be using this account:** \_\_\_\_\_

**E-Mail:** \_\_\_\_\_

**Phone:** \_\_\_\_\_

**Fax:** \_\_\_\_\_

**Signature:** \_\_\_\_\_

**Billing Address:** \_\_\_\_\_

**City, State, Zip:** \_\_\_\_\_

## FOR OFFICE USE ONLY

**Institution Account ID\*** \_\_\_\_\_

\*Assigned by Signature Genomics

## CPT CODES

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### Signature Microarrays

TEST	SPECIMEN TYPE	CPT CODES
<b>SignatureChipWG™ or SignatureChipOS™</b>	Peripheral Blood	88386(x6), 83891
	DNA	88386(x6)
	Cultured Cells	88386(x6), 83891, 88241
	Direct Amniotic Fluid	88386(x6), 83891, 88235
	Tissue, Skin, or Products of Conception (POC)	88286(x6), 83891, 88233
<b>Signature PrenatalChip®</b>	DNA	88386(x4)
	Cultured Cells	88386(x4), 83891, 88241
	Direct Amniotic Fluid	88386(x4), 83891, 88235
	Tissue, Skin, or Products of Conception (POC)	88286(x6), 83891, 88233

### Signature FISH

TEST	SPECIMEN TYPE	CPT CODES
<b>FISH (1 locus)</b>	Peripheral Blood	88230, 88271(x2)*, 88283 88273(x2)*, 88291
	Tissue, Skin, or Products of Conception (POC)	88233, 88271(x2)*, 88283 88273(x2)*, 88291
	Amniotic Fluid	88235, 88271(x2)*, 88283 88273(x2)*, 88291
<b>FISH (2 loci)</b>	Peripheral Blood	88230, 88271(x4)*, 88283(x2) 88273(x4)*, 88291(x2)
	Tissue, Skin, or Products of Conception (POC)	88233, 88271(x4)*, 88283(x2) 88273(x4)*, 88291(x2)
	Amniotic Fluid	88235, 88271(x4)*, 88283(x2) 88273(x4)*, 88291(x2)

\* Includes control FISH probe.

### Signature Karyotype

TEST	SPECIMEN TYPE	CPT CODES
<b>G-Banded Karyotype</b>	Peripheral Blood	88230(x2), 88262, 88291
	Tissue, Skin, Products of Conception (POC)	88233(x2), 88262, 88291
	Amniotic Fluid	88235(x2), 88262, 88291

### Other

TEST	CPT CODES
<b>MCC</b>	83900, 83901(x14), 83912, 83909, 83890
<b>Fragile X</b>	83891, 83894(x2), 83892(x2), 83898, 83912, 83896, 83897
<b>AFP</b>	82106
<b>ACHÉ</b>	82013

# SIGNATURE PRENATALCHIP® DISORDERS TESTED



Clinically recognized regions of the genome  
assayed by the Signature PrenatalChip®(V3.1)

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The Signature PrenatalChip may identify copy number changes associated with genetic conditions, including adult-onset conditions and/or cancer predisposition syndromes, that are potentially unrelated to the current clinical findings but that may become apparent later in life. In addition, if there is a family history of a known or suspected genetic condition unrelated to the reason for testing, please contact the laboratory to discuss prior to sample submission.

CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION	CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION
1p36 Microdeletion *	607872	Multiple	1p36	14q11.2 Microdeletion		Multiple	14q11.2
1q21.1 Microdeletion with susceptibility for thrombocytopenia-absent radius (TAR)	274000	Multiple	1q21.1	14q22-q23 Microdeletion	600037	Multiple	14q22-q23
1q21.1 Microdeletion with susceptibility to mental retardation, autism, or congenital anomalies *	612474	Multiple ACP6 candidate GJA5 candidate GJA8 candidate	1q21.1	15q11-q13 Microduplication *	608636	Multiple	15q11-q13
1q41-q42 Microdeletion/Fryns	229850	Multiple DISP1 candidate	1q41	15q13.1 Deletion including oculocutaneous albinism	203200	OCA2	15q13.1
1q44 Microdeletion		Multiple AKT3 candidate	1q44	15q13.3 Microdeletion	612001	Multiple CHRNA7 candidate	15q13.3
2p15-p16.1 Microdeletion		Multiple	2p15-p16.1	15q24.1-q24.3 Microdeletion		Multiple	15q24.1-q24.3
2p21 Deletion including hypotonia-cystinuria and homozygous microdeletions	606407	Multiple	2p21	16p11.2 Microdeletion *	611913	Multiple	16p11.2
2q32.2-q33 Microdeletion	119540	Multiple SATB2 candidate	2q33.1	16p11.2-p12.2 Microdeletion *		Multiple	16p11.2-p12.2
3q29 Microdeletion	609425	Multiple	3q29	16p13.3 Deletion including tuberous sclerosis 2, contiguous gene deletion including polycystic kidney 1	191100	TSC2	16p13.3
5q22 Microdeletion/Gardner/Familial adenomatous polyposis with MR	175100	APC	5q22.2	16p13.1 Microdeletion predisposing to autism and/or mental retardation *		Multiple	16p13.1
6p25.3 Microdeletion		Multiple	6p25.3	16p13.3 Microdeletion/Severe Rubinstein-Taybi	610543	CREBBP DNASE1	16p13.3
6q24.3 Microdeletion		Multiple	6q24.3	16q11.2-q12.2 Microdeletion/Townes-Brocks 1	107480	Multiple SALL1 candidate ZNF423 candidate	16q11.2-q12.2
7p13 Deletion including cerebral cavernous malformations, type 2	603284	CCM2	7p13	17p13.3 Deletion including nephropathic cystinosis	219800	CTNS	17p13.3
7q11.23 Microduplication *	609757	Multiple	7q11.23	17q21.3 Microdeletion *	610443	Multiple MAPT candidate	17q21.3
8p23.1 Microdeletion *		Multiple GATA4 candidate	8p23.1	22q11.2 Distal microdeletion *	611867	Multiple	22q11.2
9q22.32-q22.33 Microdeletion		Multiple TGFBFR1 candidate	9q22.33	22q11.21 Microduplication *	608363	Multiple	22q11.21
9q34 Microdeletion *	610253	Multiple EHMT1 candidate	9q34.3	22q13.3 Microdeletion *	606232	Multiple ARSA candidate SHANK3 candidate	22q13.3
9q34.13 Deletion including tuberous sclerosis 1	191100	TSC1	9q34.13	Xpter-p22.3 & Ypter-p11.32 Deletion including Langer mesomelic dysplasia, Leri-Weill dyschondrosteosis, & X-linked idiopathic short stature	249700/ 127300/ 300582	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
10q22.3-q23.31 Microdeletion		Multiple	10q22.3-q23.31	Xp11.22-linked mental retardation *		Multiple HSD17B10 candidate HUWE1 candidate	Xp11.22
11q13.3 Deletion including oto-dental	166750	FGF3	11q13.3	Xp11.23 deletions including focal dermal hypoplasia/Goltz	305600	PORCN	Xp11.23
12q14.1-q15 Microdeletion		Multiple LEMD3 candidate GRIP1 candidate	12q14.3	Xp11.3 Microdeletion	300578	Multiple RP2 candidate ZNF674 candidate	Xp11.3
12q24.21-q24.23 Microduplication *		Multiple	12q24.21-q24.23				

CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION	CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION
Xp11.4-p21.2 Contiguous gene deletion	314850	Multiple	Xp21.1	Down syndrome critical region (DSCR) *	602917	Multiple	21q22.13
Xq28 Contiguous gene deletion		L1CAM AVPR2	Xq28	Feingold	164280	MYCN	2p24.3
Adrenal hypoplasia congenita (AHC)	300200	NR0B1	Xp21.2	FMR1 Microdeletion	300624	FMR1	Xq27.3
Alagille	118450	JAG1	20p12.2	Glycerol kinase deficiency (GKD)	300474	GK	Xp21.2
Albright hereditary osteodystrophy-like/Brachydactyly-MR	600430	Multiple	2q37.3	Gorlin-Goltz/Basal cell nevus/Holoprosencephaly 7	109400/ 610828	PTCH1	9q22.32
Alpha thalassemia mental retardation (ATR-16)	141750	HBA1 HBA2	16pter- p13.3	Greig cephalopolysyndactyly	175700	GLI3	7p14.1
Androgen insensitivity	300068	AR	Xq12	Hemophilia A	306700	F8	Xq28
Angelman	105830	UBE3A	15q11.2	Hemophilia B	306900	F9	Xq27.1
Aniridia II	106210	PAX6	11p13	Hereditary hemorrhagic telangiectasia, type 2	600376	ACVRL1	12q13.13
Atrial septal defect (ASD) with atrioventricular conduction defects	108900	NKX2-5	5q35.2	Holoprosencephaly 2	157170	SIX3	2p21
Beckwith-Wiedemann, IGF2-related *	130650	IGF2	11p15.5	Holoprosencephaly 3	142945	SHH	7q36.3
Blepharophimosis, ptosis, epicanthus inversus (BPE)	110100	FOXL2	3q22.3	Holoprosencephaly 4	142946	TGIF1	18p11.31
Boston-type craniosynostosis/Parietal foramina 1 *	604757	MSX2	5q35.2	Holoprosencephaly 5	609637	ZIC2	13q32.3
Branchio-oto-renal (BOR)/Melnick-Fraser/Oto-facio-cervical (OFC)	113650/ 166780	EYA1	8q13.3	Holoprosencephaly 8	609408	Multiple	14q13.1- q13.2
Campomelic dysplasia (CMPD)	114290	SOX9	17q24.3	Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	146255	GATA3	10p14
Cat-eye *	115470	Multiple	22q11.1	Infantile hyperinsulinism with enteropathy & deafness	606528	USH1C ABCC8	11p15.1
Cerebellar hypoplasia, VLDLR-related/Hutterite dysequilibrium	224050	VLDLR	9p24.2	Infantile spasms, MAGI2-related	606382	MAGI2	7q21.11
Cerebral cavernous malformations, type 1 (CCM1)	116860	KRIT1	7q21.2	Jacobsen/11q terminal deletion disorder	147791	Multiple	11q23- qter
CHARGE	214800	CHD7	8q12.2	Joubert 4	609583	NPHP1	2q13
Choroideremia	303100	CHM	Xq21.2	Juvenile polyposis (JPS), BMPR1A-related	174900	BMPR1A	10q23.2
Cleidocranial dysplasia (CCD)	119600	RUNX2	6p12.3	Juvenile polyposis syndrome (JPS), SMAD4-related	174900	SMAD4	18q21.2
Congenital diaphragmatic hernia (CDH)	142340	CHD2 NR2F2	15q26.1 15q26.2	Kallmann 1	308700	KAL1	Xp22.31
Congenital diaphragmatic hernia 2 (CDH2) *	222400	GATA4	8p23.1	Langer-Giedion	150230	TRPS1 EXT1	8q23.3 8q24.11
Craniofrontonasal	304110	EFNB1	Xq13.1	Li-Fraumeni 1 (LFS)	151623	TP53	17p13.1
Cri-du-Chat	123450	Multiple	5p15.2	Lissencephaly 1	607432	PAFAH1B1 (LIS1)	17p13.3
Currarino	176450	MNX1	7q36.3	Lowe	309000	OCRL	Xq25
Dandy-Walker malformation (DWM)	220200	ZIC1 ZIC4	3q24	Marfan 1 (MFS1)	154700	FBN1	15q21.1
DiGeorge/Velocardiofacial (VCF)	188400	HIRA TBX1	22q11.21	Microphthalmia 3	206900	SOX2	3q26.33
DiGeorge 2	601362	Multiple	10p14	Microphthalmia 7 with linear skin defects	309801	Multiple	Xp22.2
Dosage-sensitive sex reversal *	300018	NR0B1	Xp21.2	Miller-Dieker	247200	PAFAH1B1 (LIS1)	17p13.3
				Mohr-Tranebjaerg	304700	TIMM8A	Xq22.1
				Mowat-Wilson *	235730	ZEB2	2q22.3

CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION	CONDITION	OMIM#	GENE(S)/LOCUS	LOCATION
Myoclonus dystonia	159900	SGCE	7q21.3	Split-hand/foot malformation 3 (SHFM3) *	600095	FBXW4	10q24.32
Nablis mask-like facial	608156	Multiple	8q21.3-q22.1	Split-hand/foot malformation 5 (SHFM5)	606708	DLX1 DLX2	2q31.1
Nail-patella (NPS)	161200	LMX1B	9q33.3	SRY dosage abnormalities	278850/ 306100	SRY	Yp11.31
Nephronophthisis 1	256100	NPHP1	2q13	Steroid sulfatase deficiency	308100	STS	Xp22.31
Neurofibromatosis 1 (NF1)/MR	162200	NF1	17q11.2	Synpolydactyly/Syndactyly II	186000	HOXD gene cluster	2q31.1
Neurofibromatosis 2 (NF2)	101000	NF2	22q12.2	Trichorhinophalangeal 1	190350	TRPS1	8q23.3
Neurosensory deafness, autosomal recessive (DFNB1)	220290	GJB6	13q12.11	Ulnar-mammary	181450	TBX3	12q24.21
NFIA Haploinsufficiency	600727	NFIA	1p31.3	Van der Woude	119300	IRF6	1q32.2
Norrie	310600	NDP	Xp11.3	von Hippel-Lindau	193300	VHL	3p25.3
Okhiro	607323	SALL4	20q13.2	Waardenburg I	193500	PAX3	2q36.1
Opitz	300000	MID1	Xp22.2	Waardenburg IIA	193510	MITF	3p14.1
Ornithine transcarbamylase deficiency (OTC)	311250	OTC	Xp11.4	WAGR	194072	PAX6 WT1	11p13
Pallister-Killian *	601803	Multiple	12p	Williams-Beuren	194050	ELN	7q11.23
Pelizaeus-Merzbacher *	312080	PLP1	Xq22.2	Wilms Tumor 1	194070	WT1	11p13
Pitt-Hopkins	610954	TCF4	18q21.1	Wolf-Hirschhorn	194190	Multiple	4p16.3
Potocki-Lupski/17p11.2 Microduplication *	610883	Multiple	17p11.2	X-linked agammaglobulinemia	300755	BTK	Xq22.1
Potocki-Shaffer	601224	EXT2 ALX4	11p11.2	X-linked Alport (ATS)	301050	COL4A5	Xq22.3
Prader-Willi (PWS)	176270	SNRPN	15q11.2	X-linked heterotaxy	306955	ZIC3	Xq26.3
Prader-Willi-like phenotype	176270	SIM1	6q16.3	X-linked infantile spasms, CDKL5-related	300672	CDKL5	Xp22.13
PTEN hamartoma tumor/Bannayan-Riley-Ruvalcaba (BRRS)/Cowden	158350/ 153480	PTEN	10q23.31	X-linked lymphoproliferative syndrome (XLP)	308240	SH2D1A	Xq25
Renal cysts and diabetes (RCAD) *	137920	HNF1B	17q12	X-linked mental retardation 21	300143	IL1RAPL1	Xp21.3
Retinoblastoma/MR	180200	RB1	13q14.2	X-linked mental retardation with isolated growth hormone deficiency *	300123	SOX3	Xq27.1
Rieger 1 (RIEG1)	180500	PITX2	4q25	X-linked mental retardation with microcephaly & disproportionate pontine and cerebellar hypoplasia	300172	CASK	Xp11.4
Rubinstein-Taybi (RTS)	180849	CREBBP	16p13.3	XX male	278850	SRY	Yp11.31
Saethre-Chotzen	101400	TWIST1	7p21.1	XY gonadal dysgenesis	306100	SRY	Yp11.31
Severe myoclonic epilepsy of infancy (SMEI)	607208	SCN1A	2q24.3	All 41 unique subtelomeric regions	Multiple		41 sites
Sex reversal, autosomal dominant 2 (SRA2)	154230	Multiple	9p24.3	All 43 unique pericentromeric regions/marker chromosomes	Multiple		43 sites
Simpson-Golabi-Behmel (SGBS)	312870	GPC3	Xq26.2	Aneuploidy for 24 chromosomes	Multiple		24
Smith-Magenis (SMS)	182290	RAI1	17p11.2				
Sotos	117550	NSD1	5q35.3				
Speech & language disorder 1	602081	FOXP2	7q31.1				
Split-hand/foot malformation 1 (SHFM1)	183600	SHFM1	7q21.3				

\* Duplications of these regions are associated with a syndrome/clinical phenotype.