Top 10 Genetics Resources for Pediatric Primary Care Providers

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Associate Professor of Pediatrics
UMDNJ – New Jersey Medical School

Thursday, October 25
12:00 - 12:30pm Central

Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute
Faculty

- Beth A Pletcher, MD, FACMG, FAAP
  - Associate Professor of Pediatrics at the University of Medicine and Dentistry – New Jersey Medical School
  - Immediate past Chair of the American Academy of Pediatrics Committee of Pediatric Workforce
  - Co-Director of The Neurofibromatosis Center of New Jersey
  - Serves on the University Hospital Bioethics Committee
Dr. Pletcher has no financial relationships or conflicts of interest to disclose relevant to this presentation.
Learning Objectives

Following this presentation, participants should be able to:

• Access and utilize online genetic resources to create a differential diagnosis for patients presenting with features of a genetic disorder
• Access and utilize online genetic resources developed for professionals about specific genetic conditions to improve patient care
• Recommend family support and online genetic resources for families who have a child with a specific genetic condition
Top Ten Resources

- AAP Committee on Genetics Health Supervision Guidelines - www.aap.org/visit/cmte18.htm
- ACMG Newborn Screening ACT Sheets – www.acmg.net/resources/policies/ACT/condition-analyte-links.htm
- Genetic Alliance – http://geneticalliance.org
- Unique rare chromosome group – www.rarechromo.org
- Genetics and rare conditions – www.kumc.edu/gec/support
- National Organization of Rare Disorders – www.rarediseases.org
- NY Online Access to Health – www.noah-health.org
OMIM

- Excellent resource for differential diagnoses
- Especially good for individuals with multiple congenital anomalies or with combinations of unusual facial features, cognitive delays and/or a birth defect
- Key issues
  - Need to find less common or most unusual clinical feature(s)
  - Input two or three critical features
  - Prioritize the possible diagnoses
Case #1

- Full term infant in the nursery with:
  - Microcephaly
  - Microophthalmia
  - Diaphragmatic hernia

- OMIM search uncovered 19 potential "matches" – one was a duplicate diagnosis so 18 real possible ones

- Only 3 had all three clinical features
OMIM Search Process

• Go to site
• Input two or three key clinical findings
• Hit search
• A list of possible conditions appear
• Click on each individual condition
• On upper right side click on Table of Contents (this is critical)
• Drop-down menu - select “Clinical Synopsis”
OMIM Search Results

- CHROMOSOME 1q41-q42 DELETION SYNDROME (C)
- FOCAL DERMAL HYPOPLASIA; FDH (XLD)
- CHROMOSOME 15q24 DELETION SYNDROME (C)
- COFFIN-SIRIS SYNDROME; CSS (AR)
- CORNELIA DE LANGE SYNDROME 1; CDLS1 (AD)
- PANCREATIC AGENESIS AND CONGENITAL HEART DEFECTS; PACHD (AD)
- EMANUEL SYNDROME (C)
- MICROPHTHALMIA, SYNDROMIC 7; MCOPS7 (XLD)
- KABUKI SYNDROME 1; KABUK1 (AD)
- AMINOPTERIN SYNDROME SINE AMINOPTERIN; ASSA (T)
- WOLF-HIRSCHHORN SYNDROME; WHS (C)
- MARDEN-WALKER SYNDROME (AR)
- CHROMOSOME 16p11.2 DELETION SYNDROME, 593-KB (C)
- BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS; BPES (AD)
- BOHRING-OPITZ SYNDROME; BOPS (AD)
- RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1; RCDP1 (AR)
- CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IB; ARCL1B (AR)
- C SYNDROME (AR)
<table>
<thead>
<tr>
<th>CATEGORY</th>
<th>SUBCATEGORY</th>
<th>FEATURES</th>
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<tbody>
<tr>
<td>Inheritance</td>
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<td>Isolated cases</td>
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<td>Growth</td>
<td>Height</td>
<td>Short stature</td>
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<td>Head and Neck</td>
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<td>Frontal bossing</td>
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<td>Face</td>
<td>Coarse facies</td>
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<td>Eyes</td>
<td>Dysmorphic features</td>
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<td>Deep-set eyes</td>
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<td>Hypotelorism</td>
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<td>Upslanting palpebral fissures</td>
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<td>Anteverted nares</td>
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<td>Mouth</td>
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<td>Respiratory</td>
<td>Lung</td>
<td>Lung hypoplasia in those with diaphragmatic hernia</td>
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<tr>
<td>Chest</td>
<td>Diaphragm</td>
<td>Diaphragmatic hernia</td>
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<td>Genitourinary</td>
<td>Internal Genitalia (Male)</td>
<td>Cryptorchidism</td>
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<td>Skeletal</td>
<td>Feet</td>
<td>Club feet</td>
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<td>Neurologic</td>
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<td>Developmental delay</td>
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<td>Mental retardation</td>
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<td>Seizures</td>
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<td>Holoprosencephaly</td>
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<td>Miscellaneous</td>
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<td>Highly variable phenotype</td>
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<td>Midline defects</td>
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<td>Contiguous gene deletion syndrome</td>
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<td>Molecular Basis</td>
<td>-</td>
<td>Caused by deletion (1.7Mb) of 1q41-q42</td>
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Creation Date: Cassandra L. Kniffin : 11/19/2010
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<tr>
<td>Face</td>
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<td>Facial asymmetry</td>
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<td></td>
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<td>Pointed chin</td>
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<td>Protruding, simple ears</td>
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<td>Low-set ears</td>
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<td>Ears</td>
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<td>Narrow auditory canals</td>
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<td>Hearing loss, mixed</td>
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<td>Strabismus</td>
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<td>Iris coloboma</td>
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<td>Aniridia</td>
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<td>Eyes</td>
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<td>Microphthalmia (15%)</td>
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<td>Anophthalmia</td>
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<td>Choroidoretinal coloboma</td>
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<td>Ectopia lentis (6%)</td>
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<td>Aniridia (3%)</td>
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<td>Optic atrophy</td>
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<td>Nystagmus</td>
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<td>Decreased visual acuity</td>
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<td>Nose</td>
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<td>Broad nasal tip</td>
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<td></td>
<td>Notched nasal alae</td>
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<td>Mouth</td>
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<td>Papillomas (lip, gingiva)</td>
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<td>Cleft lip</td>
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<td>Cleft palate</td>
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<td>Hypodontia</td>
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<td>Oligodontia</td>
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<td>Delayed eruption</td>
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<td>Malocclusion</td>
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<td>Notched incisors</td>
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<td>Papillomatosis</td>
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<td>Midclavicular aplasia</td>
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<td>Rib hypoplasia</td>
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<td>Asymmetric breast</td>
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<td>Nipple hypoplasia</td>
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<tr>
<td>Diaphragm</td>
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# CHROMOSOME 15q24 DELETION SYNDROME

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<td></td>
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<td>Obesity (less common)</td>
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<td>Other</td>
<td>Intrauterine growth retardation</td>
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<td>Poor postnatal growth</td>
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<tr>
<td>Head and Neck</td>
<td>Head</td>
<td>High forehead</td>
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<td>Microcephaly</td>
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<td></td>
<td>Face</td>
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<td>Smooth philtrum</td>
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<td>Microretrognathia</td>
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<td>Facial asymmetry</td>
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<td>Long face</td>
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<td>Ears</td>
<td>Large ears</td>
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<td>Cupped ears</td>
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<td>Downsloping palpebral fissures</td>
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<td>Eyes</td>
<td>Strabismus</td>
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<td>Microphthalmia, mild</td>
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<td>Epicanthal folds</td>
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<td>Hypopigmentation of the iris</td>
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<td>Nose</td>
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<td>Broad nasal bridge</td>
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<td>Flaring of nasal alae</td>
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<td>Wide, short nose</td>
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<td>Full lower lip</td>
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<td></td>
<td>Mouth</td>
<td>High arched palate</td>
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<td>Open mouth</td>
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<td></td>
<td>Diaphragm</td>
<td>Diaphragmatic hernia (less common)</td>
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# Coffin-Siris Syndrome; CSS

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<td>Postnatal growth retardation</td>
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<td><strong>Microcephaly</strong></td>
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<td>Face</td>
<td>Coarse facies</td>
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<td>Facial hypertrichosis</td>
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<td>Ears</td>
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<td>Ptosis</td>
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<td>Myopia</td>
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<td><strong>NO Micro-Ophthalmia</strong></td>
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<td>Flat nasal bridge</td>
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<td>Broad nasal tip</td>
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<td>Choanal atresia</td>
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<td>Wide mouth</td>
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<td>Full lips</td>
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<td>Mouth</td>
<td>Cleft palate</td>
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<td>Teeth</td>
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<td>Heart</td>
<td>Ventricular septal defect</td>
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<td>Atrial septal defect</td>
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<td></td>
<td>Vascular</td>
<td>Tetralogy of Fallot</td>
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<td></td>
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<td>Patent ductus arteriosus</td>
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<tr>
<td>Chest</td>
<td>Ribs, Sternum, Clavicles and Scapulae</td>
<td>Short sternum</td>
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<td>Diaphragm</td>
<td><strong>Diaphragmatic hernia</strong></td>
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Case #2

- Full term infant in the nursery with unusual facial features and a ventricular septal defect
- Input = small ears, epicanthal folds and a ventricular septal defect
- The OMIM search uncovered 41 potential diagnoses, but Trisomy 21 was not one of them! (EF & VSD = 63) (EF & cardiac defect = 55 with Tri 21 finally appearing!)
- Too much non-specific information
Case #3

• Adult patient with a history of colorectal cancer at a young age (35 years)
• In OMIM search, input “colorectal cancer”
• Result—300 potential diagnoses including individual genes without a syndrome associated with it
• Too little non-specific information
Gene Tests/ Gene Reviews

- [www.geneclinics.org](http://www.geneclinics.org)
- As of 10/17/12 this site has:
  - 569 GeneReviews
  - 1109 Clinics
  - 626 Registered Laboratories
  - 2809 Diseases (2561 Clinical & 248 Research)
- Key resources include
  - GeneReviews
  - Lab Directory
  - Clinic Directory
  - Educational Materials
GeneTests GeneReviews

GeneReviews are expert-authored, peer-reviewed, current disease descriptions that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions. Read more...

Search GeneReviews and Laboratory Directory

Enter a search term in any field above and select applicable Go button. Or click here to reset all search fields: Clear

Browse GeneReviews

All Titles
All Overviews
All Authors
Typical Search for GeneReviews

• In Web site, type in a key word
• Must be spelled correctly
• Not case-sensitive
• I typed in rett (for Rett syndrome caused by mutations in the MECP2 gene) and then clicked on Reviews
MECP2-Related Disorders
Includes: Classic Rett Syndrome, MECP2-Related Severe Neonatal Encephalopathy, PPM-X Syndrome
John Christodoulou, MBBS, PhD, FRACP, FFSc, FRCPA and Gladys Ho, BSc, MSc.
Author Information John Christodoulou, MBBS, PhD, FRACP, FFSc, FRCPA Professor, Pediatrics and Biochemical, Molecular, and Human Genetics Director, Western Sydney Genetics Program Children's Hospital at Westmead Sydney, New South Wales, Australia john.christodoulou@health.nsw.gov.au Gladys Ho, BSc, MSc NSW Centre for Rett Syndrome Research Western Sydney Genetics Program Children's Hospital at Westmead Sydney, New South Wales, Australia gladys.ho@health.nsw.gov.au Initial Posting: October 3, 2001; Last Update: June 28, 2012.
Summary
Go to:
Disease characteristics. MECP2-related disorders in females include classic Rett syndrome, variant Rett syndrome, and mild learning disabilities. A MECP2 mutation in a male is presumed to most often be lethal; phenotypes in rare surviving males are primarily severe neonatal encephalopathy and manic-depressive psychosis, pyramidal signs, Parkinsonian, and macro-orchidism (PPM-X syndrome). Classic Rett syndrome, a progressive neurodevelopmental disorder primarily affecting girls, is characterized by apparently normal psychomotor development during the first six to 18 months of life, followed by a short period of developmental stagnation, then rapid regression in language and motor skills, followed by long-term stability. During the phase of rapid regression, repetitive, stereotypic hand movements replace purposeful hand use. Additional findings include fits of screaming and inconsolable crying, autistic features, panic-like attacks, bruxism, episodic apnea and/or hyperpnea, gait ataxia and apraxia, tremors, seizures, and acquired microcephaly.
Gene Tests/Gene Reviews

• Full Rett/MECP2 review is 23 pages long
• Has sections as below:
  – Summary (brief description, diagnosis, management and genetic counseling)
  – Clinical diagnosis and diagnostic criteria
  – Molecular genetic testing with pick-up rate and links to labs
  – Testing strategies
  – Detailed clinical information and natural history
  – Differential diagnosis
  – Management
  – Registries
  – Genetic counseling
  – Resources for families
GeneTests - Finding Labs

• Look up a condition by name
• Click on “testing”
• Retrieve a list of all the labs that are offering testing for this condition in and outside the US
• View a table outlining specific testing offered (prenatal vs. postnatal, complete or targeted sequencing versus specific mutations)
• Click on an individual lab to get the name of the lab contact with phone number, fax and email address
AAP Committee on Genetics

- [www.aap.org/visit/cmte18.htm](http://www.aap.org/visit/cmte18.htm)
- Physician-friendly health supervision guidelines including prenatal advice, recommended screening protocols and in some cases disorder specific growth curves. Would recommend keeping a copy of this in your patient’s chart or scan it into your EMR.

Covers common genetic topics including:

- Achondroplasia
- Down syndrome
- Fragile X syndrome
- Marfan syndrome
- Neurofibromatosis
- Prader-Willi syndrome
- Sickle cell disease
- Williams syndrome

- Also contains other topics including:
  - Newborn Screening Fact Sheets
  - Congenital Adrenal Hyperplasia
  - Clinical Evaluation of the Child with Mental Retardation or Developmental Delays
Committee on Genetics

Mission

The Committee on Genetics studies and makes recommendations to the Board of Directors on recent advances in genetics and provides support to chapters on state legislative issues as they relate to genetics.

AAP Policy Documents Authored by the COG

- Clinical Criteria: Evaluation of the Child with Mental Retardation or Developmental Delay
- Congenital Anatomical Abnormalities
- Fetal Alcohol Effects: Prevention of Fetal Alcohol Syndrome
- Health Supervision for Children with Down Syndrome
- Health Supervision for Children with Fragile X Syndrome
- Health Supervision for Children with Autism Spectrum
- Health Supervision for Children with Neurofibromatosis
- Health Supervision for Children with Ehlers-Danlos Syndrome
- Health Supervision for Children with Williams Syndrome
- Auditory Brainstem Response
- Auditory Brainstem Response in Pediatric Practice
- Newborn Screening Facts Sheets: Introduction
- Newborn Screening Fact Sheets
- Update of Newborn Screening and Therapy for Congenital Hypothyroidism
- Newborn Screening Evaluation

Selected List of Other Genetics-Related AAP Policy Documents

- Ethical Issues Xanthine Oxidase Testing in Pediatrists
- Oculofacial Hereditary Syndrome: Oculofacial Syndrome
- Fetal Alcohol Syndrome and Alcohol-Related Neurodevelopmental Disorders
- Human Embryo Research
- Auditory Integration Training and Field Elicited Otoacoustic Emissions for Autism
- The Warehouse
- Behavior Modification for Special Dietary Use
- Principles and Guidelines for Early Hearing Detection and Intervention Program

Patient Education Brochures and Other Materials related to Genetics Issues

The following AAP Patient Education Brochures (in bulk quantities) and other materials are available for purchase by contacting the AAP Bookstore online or by phone at 888-222-1789:
- Newborn Hearing Screening and Your Baby
- Learning Disabilities: What Elementary School Teachers Need to Know

What is a Pediatric Geneticist?

The AAP has created a series of fact sheets that contain information about different pediatric subspecialists which parents may be referred to. The "What is a Pediatric Geneticist" fact sheet can be found here. The list of all AAP subspecialty fact sheets can be found here.
eMedicine

- Hosted by Medscape LLC (from WebMD)
- Summary of many medical and genetic conditions
- Has undergone revisions over the past years and some of the authors are more junior people or lack specific expertise and sometimes not all of the information is totally correct
- Benefits—great pictures and radiographs
- Currently is being reformatted so topics match each other in style and content
Pediatrics: Genetics and Metabolic Disease Articles

- Achondrogenesis
- Aicardi Syndrome
- Apert Syndrome
- Arthrogryposis
- Ceroid Lipofuscinoses
- CHARGE Syndrome
- Chromosomal Breakage Syndromes
- Cornelia De Lange Syndrome
- Cri du chat Syndrome
- Danon Disease
- Down Syndrome
- Ellis-van Creveld Syndrome
- Fragile X Syndrome
- Genetics of Achondroplasia
- Genetics of Asphyxiating Thoracic Dystrophy (Jeune Syndrome)
- Genetics of Cockayne Syndrome
- Genetics of Crouzon Syndrome
- Genetics of Ehlers-Danlos Syndrome
- Genetics of Fabry Disease
- Genetics of Hidrapl-Erb-Flaxa-Weber Syndrome
- Genetics of Marfan Syndrome
- Genetics of Nail-Patella Syndrome
- Genetics of Neurofibromatosis
- Genetics of Niemann-Pick Disease
- Genetics of Osteogenesis Imperfecta
- Genetics of Proteus Syndrome
- Genetics of Rett Syndrome
- Genetics of Sjogren-Larsson Syndrome
- Genetics of Tuberous Sclerosis
- Genetics of Waardenburg Syndrome
- Holoprosencephaly
- Klippel-Feil Syndrome
- Klinefelter Syndrome
- Mandibulofacial Dysostosis (Treacher Collins Syndrome)
- Meckel-Gruber Syndrome
ACMG Newborn Screening ACT Sheets

- [www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm)
- American College of Medical Genetics and Genomics (ACMG)
- Click on “Newborn Screening ACT Sheets and Confirmatory Algorithms”
- Search for the condition of interest (CAH, MSUD, PKU, methylmalonic acidemia, etc...)
- Look at the ACT sheet – very brief description of the condition and **what you need to do**
- Look at the algorithm to see a concise stepwise plan of action
ACMG ACT Sheets

The following table describes the interrelationships between the conditions screened in newborn screening laboratories and the markers (analytes) used for screening. For each marker(s), there is 1) an ACT (ACT) sheet that describes the long-term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow-up of the infant that has screened positive, and 2) an algorithm that presents an overview of the basic steps involved in determining the final diagnosis in the infant. By clicking on either the ACT sheet or Algorithm for a particular condition in the column labeled "Links," you will be linked to the corresponding materials.

The algorithms were developed by a work group that included experts in the various specialties and conditions involved in newborn screening for endocrine, hematological, genetic and metabolic diseases. The first page of the ACT sheets includes information that was developed by the experts. It includes links to informational resources to allow the health professional to obtain additional information, if needed. The second page of the ACT sheet includes links to web sites that allow one to identify subspecialists for consultation and referral for the condition(s) described in the ACT sheet. It is supplemented with links to your State Newborn Screening Program and is available for the inclusion of additional health professionals who may be consulted in your area.

All materials were approved by the Board of Directors of the American College of Medical Genetics and Genomics (ACMG) on December 26, 2009. The materials will be maintained by ACMG over time and additional materials, added as new conditions are introduced into newborn screening programs. The project was partially funded through grant U22MC039197 from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services.

- Newborn Screening ACT Sheets and Confirmatory Algorithms
- Transition ACT Sheets and Confirmatory Algorithms
- Family History ACT Sheets and Confirmatory Algorithms
- Carrier ACT Sheets and Confirmatory Algorithms
- About the ACMG ACT Sheets and Confirmatory Algorithms
Genetic Alliance

- [http://geneticalliance.org](http://geneticalliance.org)
- Directory of genetic support groups
- “Disease InfoSearch” on the left side
  - Disorders are listed alphabetically
  - Find your condition – warning the lists are long so you may need to scan multiple pages (for instance Cornelia de Lange is on page 19 of the “C’s”)
  - The links are all there to the support groups including brochures and contact names and numbers
  - Well-organized and frequently updated
- Baby’s First Test—[www.babysfirsttest.org](http://www.babysfirsttest.org)
Unique Rare Chromosome Group

- www.rarechromo.org
- Site is for chromosome abnormalities only
- Go to “Information” on the left side
- Then for an overview click on “Chromosomes and Disorders”
- For specific cytogenetic variations instead click on “Disorders Leaflets”
- Search for your specific condition
- Some leaflets are in other languages including German, Spanish, French and Italian
Welcome to Unique

Unique is a source of information and support to families and individuals affected by any rare chromosome disorder and to the professionals who work with them. Unique is a UK-based charity but welcomes members worldwide. Membership of Unique is free but the group receives no government funding and is heavily reliant on donations and fundraising to continue its work. Please help us in whatever way you can.

You may have been given a diagnosis or indication of a chromosome disorder by a geneticist or other medical professional and they may have used a medical term which is unfamiliar to you. So to help you decide if Unique is the appropriate organisation for you, we thought it would be useful to describe the different categories of rare chromosome disorder. Rare chromosome disorders can be grouped as structural disorders, numerical disorders and other miscellaneous disorders.

Click on the image above to download our free information leaflets on many chromosome disorders.
Genetics and Rare Conditions

- [www.kumc.edu/gec/support](http://www.kumc.edu/gec/support)
- This site developed by the University of Kansas is amazing in its simplicity
- It is merely an alphabetical listing of rare conditions and genetic disorders
- If you click on your condition of interest you get connected to a list of support groups and sites including those in other countries and in other languages
KUMC

Genetic and Rare Conditions Site

Lay advocacy and support groups, information on genetic conditions / birth defects for professionals, educators, and individuals. National and international organizations

- Categories: Genetic Counselors and Geneticists · Children and teen sites · Advocacy · Diversity · Spanish sites · Other · Suggestions · Search

A B C D E F G H I J K L M N O P Q R S T U V W X Y Z

- Aarskog syndrome
- Achondroplasia
- Achromatopsia
- Acoustic neuroma (and benign cranial nerve tumors)
- Adrenal hyperplasia
- Adrenoleukodystrophy
- Agenesis of corpus callosum
- Alcardi syndrome
- Alagille syndrome
- Albinism (and hypopigmentation)
- Alopecia areata
- Alstrom syndrome
- Alpha-1-antitrypsin deficiency
- Ambiguous genitalia
- Androgen insensitivity syndrome(s)
- Anorchia
- Angelman syndrome
- Anophthalmia
- Apert syndrome
- Arthrogryposis (amyoplasia)
- Ataxia (Friedreich ataxia, spinocerebellar ataxias, ataxia telangiectasia, essential tremor, spastic paraplegia, other)
National Organization of Rare Disorders

- [www.rarediseases.org](http://www.rarediseases.org)
- Nice site for patients and physicians—requires practice navigating at first
- On the left look at “Patient Info Center” and your patient can – Ask a Nurse, Ask a Genetic Counselor, or find a doctor through the Physician Database
- Tool bar at the top – go to Rare Disease Info and then Rare Disease Database
- There you will find an alphabetical listing of conditions
- Each condition has a nice description including symptoms and causes as well as treatments (standard and investigational) and finally links to support organizations
NY Online Access to Health

- [www.noah-health.org](http://www.noah-health.org)
- Access to complete text of consumer friendly information – not developed by them but others
- Information can be searched for in English, Spanish or both
- It is not only a genetic site, so you need to first go to “Health Topics” then “Genetics”
- If you want to look for something in Spanish, you type in the name of the condition on the upper right side and click Spanish
Summary

• Many of these wonderful resources are the geneticists’ “best kept secrets”
• Don’t be reluctant to spend 3 to 5 minutes on a few of these sites to see what you can find
• These resources can assist you in daily practice to:
  – Initiate a diagnostic work-up
  – Respond to unexpected clinical situations – abnormal newborn screening test, prenatal fetal abnormalities, a newborn with multiple congenital anomalies or an older child or adult with a known genetic condition
  – Help families begin to understand more about their specific genetic condition and help them connect with support groups
Additional Resources for PCPs—Continuing Education

• Online CME activity—ACMG Basics: Genetics for Providers, which designates up to 6 hours of AMA PRA Category 1 Credit TM toward completion of the AMA Physician's Recognition Award.
  – [http://www.acmg.net/AM/Template.cfm?Section=CME_Activities&Template=/CM/HTMLDisplay.cfm&ContentID=3106](http://www.acmg.net/AM/Template.cfm?Section=CME_Activities&Template=/CM/HTMLDisplay.cfm&ContentID=3106)

• The AAP Committee on Genetics is putting together a resource on genetics for primary care providers, the manual, Genetics in Primary Care: A Handbook; available in spring 2013

• “Genetics in Medicine, 7th Edition” is an excellent text with basic genetics for the clinician

• Genetics in Primary Care Institute—Technical Assistance Center will be launched in January 2013 with comprehensive resources, information, and listing of continuing educational opportunities
Additional Resources for PCPs—Family History

- March of Dimes: Genetics & Your Practice
  - “Family Health and Social History”
    www.marchofdimes.com/gyponline/index.bm2

- GeneTests
  - www.genetests.org

- The American Society of Human Genetics—Health Provider Genetics Resources
  - www.ashg.org/press/healthprofessional.shtml

- Genetic Science Learning Center (Utah)
  - http://learn.genetics.utah.edu/content/health/history
## Additional Resources for PCPs—Pre/Neo/Postnatal History

<table>
<thead>
<tr>
<th>Prenatal</th>
<th>Neonatal</th>
<th>Postnatal</th>
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<tbody>
<tr>
<td><em>Smith’s Recognizable Patterns of Human Malformation</em>, 6th ed. 2006</td>
<td></td>
<td><em>Human Malformations and Related Anomalies</em> (for structural malformations)</td>
</tr>
<tr>
<td><em>Human Malformations and Related Anomalies</em>, 2nd ed. 2006</td>
<td></td>
<td>Dysmorphology databases, like POSSUM (designed for the medical geneticist)</td>
</tr>
</tbody>
</table>
Additional Resources—Newborn Screening

• Newborn Screening Clearinghouse
  – www.babysfirsttest.org *(Families and Providers)*
• Regional Genetic and Newborn Screening Collaboratives
  – http://www.nccrcg.org/AM/Template.cfm?Section=Resource_Center3
• State Genetics Program (by state) *(Families and Providers)*
  – http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm
## Additional Resources for PCPs—Genomics

<table>
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<tr>
<th>CDC</th>
<th>National Coalition for Health Professions Education in Genetics</th>
<th>National Human Genome Research Institute</th>
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Questions?
Acknowledgments

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Thank you for your participation!

For more information, please contact
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847/434-7612

www.medicalhomeinfo.org/GPCI.aspx
Time Out for Genetics

Registration is now open for

“Genetics Across the Lifespan – Putting it All Together”

Melissa Parisi, MD, PhD, FAAP, FACMG

Thursday, November 29
12:00 - 12:30pm Central

https://www2.gotomeeting.com/register/634989450