Regional Genetic Services: Past, Present & Future

March 10th, 2007

Nancy Shinno, MD-Regional Chief
Mónica Alvarado, MS-Administrator

"Well, Frank's hoping for a male and I'd like a little female. . . . But, really, we'll both be content if it just has six eyes and eight legs."
Genetic Services Vision and Mission Statements

Vision Statement:
We strive to serve as a model of excellent genetic services in the total health care system through: Compassion, Quality, Service, and Prevention.

Mission Statement:
Our mission is to provide accessible, high quality genetic services with compassion to our members through patient advocacy, diagnosis and education to:
• Optimize health care options for people affected by the challenges of a genetic condition
• Improve quality of life, health, and pregnancy outcomes for our members
• Reduce morbidity and mortality
• Empower people affected with genetic conditions or congenital disorders

Real Stories: Impact of Genetic Services on our members
• High quality and accessible services
• Focus on patient empowerment and preventive care
• Compassionate, Caring and Dedicated
• Support healthy choices for our members and their families throughout their lives
• Exemplify the diversity and excellence of resources and services offered by SCPMG
GENETICS IN THE 20TH CENTURY
1950’s - 1960’s

• Correct number of human chromosomes confirmed
• Down Syndrome found to be caused by extra chromosome
• Molecular basis of sickle cell disease delineated
• Watson and Crick described DNA
• Cytogenetic Dx of Patau, Edward, Cri du Chat syndromes
• Philadelphia chromosome found in leukemia
• Change of diet found to prevent mental retardation in children with Phenylketonuria

• SCPMG founded, key developments:
  • Harold Bass, MD, joined SCPMG at PC
  • David Weinstein, MD, joined SCPMG at HC
  • Established cytogenetics lab
  • Newborn Screening (NBS) started to test for Phenylketonuria (PKU)

GENETICS IN THE 20TH CENTURY
1970’s

• Chromosome banding was perfected to allow diagnosis of specific chromosomal abnormalities
• Population Screening for Tay Sachs was started
• Amniocentesis for prenatal diagnosis widely available

SCPMG developments:
• Dr. Bass started the Tay Sachs Screening Program
• Diane Broome, MD, & Nancy Shinno, MD, joined SCPMG at Los Angeles and WLA
• George Lewis, MD, established Craniofacial Services
• Cytogenetics Lab/Genetic Testing reestablished at the Regional Reference Laboratories
GENETICS IN THE 20TH CENTURY

1980’s

• DNA Technology expanded rapidly: restriction enzymes, recombinant DNA and polymerase chain reaction
• Prenatal Screening: Down Syndrome, Neural Tube Defects and Hemoglobinopathies
• Prenatal Diagnosis: High level ultrasound, chorionic villus sampling, umbilical cord blood sampling
• Increasing Genetic studies used in Oncology Protocols
• California’s AFP testing program began in 1986

SCPMG developments:
• Dr. Bass became Coordinator of Genetic Services
• Dr. Broome organized Prenatal AFP Program and hired our first genetic counselor
• Mehdi Jamehdor, MD, joined SCPMG & expanded the Genetic Testing Lab
• Drs. Shinno & Elaine Smith organized prenatal screening for hemoglobinopathies
• Genetic counselors were hired at all service areas
• Ronald Reinsch, MD, started chorionic villus sampling
• Newborn Screening expanded to include galactosemia & hypothyroidism
• Drs Bass & Broome began pre-symptomatic testing for Huntington disease

GENETICS IN THE 20TH CENTURY

1990’s

• DNA Technology explodes, trinucleotide repeats found in Fragile X Syndrome, Huntington’s and other disorders
• Fluorescent In Situ Hybridization (FISH) able to detect submicroscopic chromosome deletion, e.g. Di George syndrome
• Screening for Hereditary Breast, Colon & other Cancers became possible
• Prenatal Screening: AFP expanded
• Newborn Screening for Hemoglobinopathies started in California

SCPMG developments:
• Expansion of the Metabolic Service under direction of Dr. Rebecca Mardach
• Sickle Cell/Hemoglobinopathy Center
• Dr. Shinno became Medical Director of Craniofacial Services
• Dr. Jamehdor expanded the laboratory, helped establish an Interregional cytogenetics registry, Breast Cancer Screening Guidelines, and participated in Federation expansion
• First Cancer Genetics Symposium & Interregional Task Force for Guidelines for Hereditary Breast and Ovarian Cancer
GENETICS IN THE 21ST CENTURY

• Human Genome Project completed and raised expectations
• Use of tandem mass spectroscopy & "DNA chip"
• Pharmacogenetics & Pharmacogenomics expand
• Individualized Treatment for Cancers & other diseases e.g. Herceptin
• Detection of predisposing genes in common disorders

SCPMG developments:
• SCPMG performed first bone marrow transplant for sickle cell disease. In the future we may have less traumatic “genetic cures”
• Newborn screening for congenital deafness began in 2003
• Prenatal screening for cystic fibrosis beginning end of 2003
• Neurogenetic Service began in 2003
• 2004 Genetics became first Regional Department at SCPMG with Dr. Nancy Shinno as Regional Chief and Mónica Alvarado, MS, as Regional Administrator
• First OB-GYN Geneticist, Pathologist Geneticist and skeletal dysplasia expert joined department
• Regional Genetics active in “marketing” internally
• Demand for genetics services soars: 25% increase for genetic counselors, 41% for MDs from 2005 to 2006
• First Trimester Screening scheduled to begin in 2007

Regional Genetics in 2007

• Providers:
  - 8.40 FTE MD Geneticists
  - 25.20 FTE Genetic Counselors
  - Metabolic Clinic: Dietician & Social Worker

• Regional Genetics Services
  - Prenatal/Reproductive Genetics
  - Neonatal/Pediatric Genetics
  - Adult Genetics including Cancer & Neurogenetics
  - Genetic Screening Programs
  - Metabolic Genetics Clinic
  - Craniofacial Service
  - Genetic Testing Laboratory
Indications for Genetic Counseling: Preconception & Prenatal

- Those who have a personal or family history of an inherited disorder, developmental delay or a birth defect.
- Women who are pregnant or planning to have a child at age 35 yrs or older.
- Pregnant women whose ultrasound examinations or blood tests indicate that their pregnancy may be at increased risk for certain birth defects.
- Individuals who already have a child with mental retardation, an inherited disorder or a birth defect.

Indications for Genetic Counseling: Preconception & Prenatal Part II

- Known carriers of genetic disorders or chromosome rearrangements
- Women who have had babies who died in infancy or three or more first-trimester miscarriages
- Couples who are first cousins or other close blood relatives.
- People concerned about exposures to agents that may pose a risk to outcome of pregnancy. Common causes of concern include exposure to radiation, medications, illicit drugs or infections.
Indications for Genetic Evaluation: Pediatric

• Children born with congenital malformations
• Children with hearing loss or blindness
• Children with developmental delay, mental retardation or autism
• Children with tumors that may have a hereditary component (retinoblastoma, Wilm’s tumor, etc.)
• Children with unusual physical characteristics (unusual facial features, unusual growth, etc.)
• Children of parents with known autosomal dominant disorders (e.g., Neurofibromatosis, Marfan syndrome, Familial polyposis)

Indications for Genetic Evaluation: Adult

• Adults of reproductive age who were born with congenital malformations or genetic disorders
• Adults with hearing loss or blindness
• Adults with unusual physical characteristics (unusual facial features, unusual growth, etc.) of unknown etiology
• Individuals with a family history of adult-onset autosomal dominant conditions (e.g., Huntington disease, Inherited susceptibility to breast/ovarian cancer, etc.)
• Personal or family history of stroke or blood clot before age 50, or those with known thrombophilia mutation in the family
Family History: Minimum requirements

- Obtain family history information on at least a three-generation's family
- Ask about all individuals in both sides of the patient’s family and record pregnancy history including losses/stillbirths/neonatal deaths, age at diagnosis of significant disease, current age or age at and cause of death
- Ask about history of mental retardation/developmental delay, birth defects, known genetic disorders
- Record ethnicity and race
- Record consanguinity

Three-Generation Pedigree
Autosomal Dominant Inheritance

- Each child has 50% chance of inheriting the mutation
- No “skipped generations”
- Equally transmitted by men and women

Elements of Genetics Evaluation

- Collection & Assessment of family and medical history
- Physical exam (when indicated)
- Risk Assessment
- Education and Counseling
- Psychosocial Assessment
- Genetic Testing (when indicated)
- Process of Informed Consent
- Result Interpretation
- Management Plan
Types of Genetic Tests

- **Diagnostic Tests** are used to confirm or exclude a suspected genetic condition in symptomatic individuals of any age.
- **Carrier Tests** are used to identify healthy persons who have a genetic mutation for an autosomal or X-linked recessive disorder which puts their children at risk for having that disorder.
- **Prenatal Tests** are used to diagnose genetic conditions in the fetus. They are offered when there is an increased risk of having a child with a genetic condition.
- **Predictive Tests** are offered to asymptomatic individuals concerned about susceptibility to a genetic disorder. There are two types of predictive testing on. 1. Presymptomatic, where the eventual development of symptoms is certain, e.g. Huntington Disease. 2. Predispositional where the eventual development of symptoms is likely, but not certain, e.g. Breast and Ovarian Cancer.

Types of Genetic Tests

- **Pharmacogenetic Tests** are used to determine how an individual’s genetic makeup may affect reactions to specific drugs. They may help providers prescribe the most effective drugs with the least side effects.
- **Newborn Screening Tests** are used on newborns to determine whether they are at an increased risk of having specific genetic conditions which usually need treatment immediately. By state law, all babies are screened unless the parents specifically decline testing.
- **Preimplantation Genetic Diagnosis (PGD)** is performed on early embryos resulting from in vitro fertilization to decrease the risk of a particular genetic condition occurring in the fetus. It’s safety and efficacy is not completely determined.
Regional Genetic Testing Lab:
Top 10 Genetic tests in 2006

**NAPS**
1. Newborn Screening (TSH, 170HP, GAL, Hgb, Tandem MS)
2. Prenatal Screening (AFP, uE3, HCG)

**Molecular Genetics**
3. Cystic Fibrosis
4. Factor V
5. Factor II

**Cytogenetics**
6. Cancer Cytogenetics
7. Amniotic Fluid Cytogenetics
8. Molecular Cytogenetics (F.I.S.H.)

**Biochemical Genetics**
9. Plasma Homocysteine
10. Plasma Amino Acids

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DNA Testing Trends 1991 to 2004
Total Prenatal Genetic Counseling Encounters vs. Deliveries per Area

2006 Encounters/Deliveries

Prenatal Dx by Indication

- amniocenteses 2638, cvs 73

- XAFP, 26%
- UZ ABN, 10%
- HbSS, 0%
- REPEAT, 0%
- PG MGT, 1%
- OTHER, 1%
- MAT ANX, 2%
- HX ABN, 2%
- AGE, 58%

2006 Prenatal Diagnosis Statistics
Genetic Carrier Testing

- Carrier screening tests are optional.
- Appropriate tests should be offered to patients of particular ethnic backgrounds.
- Many carrier screening tests detect the majority of, but not all carriers. Detection rates may differ by ethnic background of the patient. Informed consent should include the fact that negative carrier tests do not completely eliminate the risk for that genetic condition in future offspring.
- Carrier screening prior to the onset of pregnancy allows more options for carrier/carryer couples (i.e. not becoming pregnant, using donor sperm from a non-carrier, prenatal diagnosis, adoption, etc).

Conditions Screened on a Genetic Basis

- Cystic Fibrosis
- Tay-Sachs Disease
- Canavan Disease
- Familial Dysautonomia
- Hemoglobinopathies
  - Sickle Cell Disease
  - Alpha- and Beta-Thalassemia
Carrier vs. Affected (Autosomal Recessive conditions)

• All genes come in pairs - one copy from father and one copy from mother
• For autosomal recessive conditions, a “carrier” is someone with a genetic mistake in just one of the copies of a given gene (and doesn’t normally show symptoms)
• People who have autosomal recessive conditions have a mutation in both copies of that gene

Carrier Screening is Needed ONLY ONCE in Life

• Carrier Screening for each condition is necessary only once in life (not once per pregnancy!)
• This is different from XAFP screening, which is required once per pregnancy
  ➢ Down syndrome and other chromosome abnormalities are normally not hereditary.
  ➢ Autosomal recessive conditions are hereditary.
Cystic Fibrosis

• Lung problems, including chronic coughing, wheezing and air trapping (asthma like symptoms).
• Pancreatic disease
• Male infertility and occasionally causes female infertility
• Most commonly seen in Caucasians
  ➢ Carrier risk: 1/25  Incidence 1/2,500

Tay-Sachs Disease

• Hexosaminidase A deficiency, causing accumulation of ganglioside in brain
• Progressive weakness, loss of motor skills, decreased attention, and increased startle response beginning at 3-6 months of age
• Death usually before 5 y/o
• Most commonly seen in Ashkenazi Jewish (AJ) population (1/30 carrier risk, occurs 1/3,600 birth)
• Also higher risk in the Creole, Cajun, French Canadian, and Moroccan Jewish populations
Canavan Disease

- Large head, lack of head control, severe hypotonia, and developmental delays by 3-5 months of age.
- Life expectancy usually in the teens.
- Most common in AJ population.
- Carrier risk: 1/57, incidence is 1/6,400 to 1/13,500 in the AJ population.

Familial Dysautonomia

- Progressive neuronal degeneration continues throughout life.
- Gastrointestinal problems, vomiting, recurrent pneumonia, and cardiovascular diseases.
- Shortened life expectancy
- Most commonly seen in the AJ population
  - Carrier risk: 1/36, incidence 1/3700
Hemoglobinopathies

• Sickle cell disease
  ➢ Commonly seen in African Americans
  ➢ Hemolysis, vascular occlusion, chronic organ dysfunction, and pain and swelling of hands.

• Beta-thalassemia
  ➢ Commonly seen in Mediterranean, Middle Eastern and some South and Central American countries
  ➢ Severe anemia and hepatosplenomegaly

Hemoglobinopathies (cont.)

• Alpha-thalassemia
  ➢ Commonly seen in the Asian (except Japanese and Korean) and African American populations.
  ➢ Hemolytic anemia, mild jaundice, and hepatosplenomegaly
  ➢ In severe case, causes hydrop fetalis

• Other hemoglobin variants
  ➢ Such as hemoglobin C and E, are seen in a specific ethnic backgrounds.
When should Patients be Referred to Genetics?

- If a patient and/or her partner receives a positive result for any screening test (during current or prior pregnancy).
- Low MCV with normal iron studies
- Abnormal hemoglobin electrophoresis result
- Family history of these genetic conditions

How Much Breast and Ovarian Cancer Is Hereditary?

Breast Cancer
- 5%–10%
- 15%–20%

Ovarian Cancer
- 5%–10%

- Sporadic
- Family clusters
- Hereditary
<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Gene</th>
<th>Cancers</th>
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<tbody>
<tr>
<td>Hereditary Breast/Ovarian</td>
<td>BRCA1</td>
<td>Breast, Ovarian, Pancreas, Others</td>
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<tr>
<td></td>
<td>BRCA2</td>
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<tr>
<td>Hereditary Non-Polyposis Colorectal Cancer (HNPCC)</td>
<td>MLH1</td>
<td>Colorectal (CRC), Endometrial, Gastric, Ovarian, Other GI, Urinary tract</td>
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<tr>
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<td></td>
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<td></td>
<td>MSH6</td>
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<tr>
<td></td>
<td>PMS2</td>
<td></td>
</tr>
<tr>
<td>Familial Adenomatous Polyposis (FAP)</td>
<td>APC</td>
<td>Colorectal, Duodenal, Thyroid, Brain</td>
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</tbody>
</table>

**Hereditary Cancer Risk: What to look for**

- **Early age of onset**
  - Usually under age 50
- **Multiple primary cancers**
  - Bilateral breast, Breast and ovarian, colon and endometrial, multiple colorectal
- **Two or more relatives on the same side of the family with the same cancer**
- **Ashkenazi Jewish Ancestry (BRCA1/2)**
- **Rare cancers**
  - Male breast cancer
BRCA1/2 Mutations Increase Risk of Breast and Ovarian Cancer
By age 30 By age 50 By age 70

<table>
<thead>
<tr>
<th></th>
<th>Breast Cancer</th>
<th>Ovarian Cancer</th>
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<tr>
<td>Population Risk</td>
<td>&lt;0.5%</td>
<td>&lt;&lt;&lt;1%</td>
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<tr>
<td>BRCA Carrier Risk</td>
<td>2% to 3%</td>
<td>&lt;&lt;1%</td>
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</table>

Clinical Features of Hereditary Non-Polyposis Colorectal Cancer (HNPCC)

- Early but variable age at CRC diagnosis (~45 years)
- Tumor site in proximal colon predominates
- Extracolonic cancers:
  - Endometrium
  - Ovary
  - Small bowel, bile duct, sebaceous skin tumors
HNPCC Increases the Risk of Colorectal Cancer

By age 50  
Population Risk 0.2%  
HNPCC Risk >25%

By age 70  
2%  
80%

Gastroenterology 1996;110:1020-7  
Int J Cancer 1999;81:214-8

Redefining What is Possible: The Future of Genetics at SCPMG

• Integration of Genetic Services with all Primary Care specialties at SCPMG
• Broader scope of practice will allow all members to benefit from the diversity of Genetic Services and technologies
• Contribute to and benefit from collaborative clinical research projects
• Create a model for the delivery of Genetic Services within KP and the community at large
Standardization/Sharing Best Practices

- **Goal**: Improving referral and follow-up time for patients with abnormal prenatal ultrasounds
  - Patient to be referred to genetics when indicated within 5 days from ultrasound
  - Patient scheduled for genetics appointment within 7 days of referral

- **Prenatal Ultrasound Abnormalities Referral to Genetics: Quality, Service, Access**
  - Abnormalities identified in Diagnostic Imaging/Radiology Dept.
  - Identify elapsed time from ultrasound exam date to date of referral to genetics
  - Identify elapsed time from referral date to genetics appointment

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**Time Span from SonO to Genetic Appt.**

<table>
<thead>
<tr>
<th>Location</th>
<th>Ref to Appt</th>
<th>SonO to Ref</th>
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<tbody>
<tr>
<td>San Diego</td>
<td>0.5</td>
<td>3</td>
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<tr>
<td>Bakersfield</td>
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<td>3.5</td>
</tr>
<tr>
<td>Pan City</td>
<td>1.5</td>
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</tr>
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<td>WLCA</td>
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<td>4.8</td>
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<tr>
<td>Orange C Bellflower</td>
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<td></td>
</tr>
<tr>
<td>Metro LA</td>
<td>1.5</td>
<td>10.6</td>
</tr>
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</table>

N=127  N=5  N=19  N=23  N=21  N=22  N=9
Average Gestational Age (in Weeks) at Post-Sonogram Genetics Appointment by Facility

Range in weeks: 21-27

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<thead>
<tr>
<th>Facility</th>
<th>Number of Weeks</th>
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<tr>
<td>South Bay (n=127)</td>
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<tr>
<td>San Diego (n=127)</td>
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<tr>
<td>Bakersfield (n=5)</td>
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<tr>
<td>Panorama City (n=19)</td>
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<tr>
<td>West LA (n=23)</td>
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</tr>
<tr>
<td>Bellflower (n=22)</td>
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<tr>
<td>Metro LA (n=9)</td>
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<tr>
<td>Baldwin Park (n=6)</td>
<td>19</td>
</tr>
<tr>
<td>Fontana (n=10)</td>
<td>18</td>
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</tbody>
</table>

Genetics Member Health Education

- Develop Genetics Member Health Education Resource directory
  - Includes websites providing patient education tools in various areas of Genetics
  - Post resource list in Genetics share-drive
  - Update as needed

- Needs assessment and development of new Genetics education materials
  - First Trimester Screening: Develop educational tool to inform patients of various screening and testing options available to them

- Develop other member educational tools/opportunities as needed
Genetics Member Health
Education

Future Plans

• Locate and/or develop more Spanish-language resources
  - Sickle cell handout
  - Create Spanish-language prenatal screening brochure, including First Trimester Screening information
• Update Cancer Genetics educational resources
• Update the “Choices in Prenatal Diagnosis-Option for Genetic Testing” brochure
  - Information on First Trimester Screening needs to be added

Genetics Multidisciplinary Clinics

• Existing clinics: craniofacial, metabolic, neurogenetic
• Needs: increase regional awareness; continue to develop and revise clinical guidelines
• Establish needs for additional clinics: renal disorders; skeletal dysplasias
Marketing Genetic Services
Genetics Marketing Initiatives

• Genetic Connection Newsletter
• Logo
• Increased visibility at KP symposia and general community
  – Presentations and exhibits
• Website visibility (intranet & internet)
• Library of genetics presentations on share drive

The Genetic Connection

With the rise of genetics becoming better defined in the practice of medicine, genetic counseling providers are increasingly involved in planning a coordinated role in many multidisciplinary clinics. Providers from the Departments of Psychiatry, surgery, obstetrics and gynecology, neurology, rheumatology, and cardiology have begun to collaborate with the genetic counseling services to develop patient care plans. The Genetic Connection is an ongoing newsletter designed to keep all members of the multidisciplinary team informed of genetic counseling activities and the use of genetics in medical practice.

Kaiser South, Santa Ana, CA: The Huntington Center for Genetic Counseling

The Huntington Center for Genetic Counseling offers comprehensive services for the diagnosis and management of genetic diseases and disorders. Services include evaluation of patients with suspected genetic conditions, family history assessment, genetic counseling, and prenatal and newborn screening services. The center also offers a genetics education program for the general public.

Regional Genetics: The Regional Genetics Program provides genetic counseling and testing services to patients and their families throughout the southern California region. Services include diagnostic testing, prenatal carrier screening, and counseling for individuals with a family history of genetic conditions. The program is funded by the state of California and is staffed by genetic counselors and other medical professionals.

Genetics & Beyond: Multidisciplinary Clinics

Genetics, in conjunction with other medical specialties, plays a critical role in the management of a variety of genetic disorders. The Regional Genetics Program offers genetic counseling and testing services to patients and their families throughout the southern California region. Services include diagnostic testing, prenatal carrier screening, and counseling for individuals with a family history of genetic conditions. The program is funded by the state of California and is staffed by genetic counselors and other medical professionals.

Inherited metabolic disease affects approximately 1 of every 10,000 children. Many of these children are identified through the State of California’s newborn screening program. Older children, adolescents, and adults of all ages are referred to the center when a metabolic disorder is suspected or diagnosed.

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SCPMG Genetics Intranet Site

Marketing Genetic Services

Genetics Marketing
Conference Exhibits

• Inform KP providers that the Department of Genetics is available in all MSAs
• Provide guidelines for Genetics referrals
• Increase awareness of Genetics at KP to the community at large
Marketing Genetic Services
Prenatal Genetics Exhibit

Marketing Genetic Services
Cancer Exhibit at KP Breast Care Symposium
### Genetics Customer Survey

**How is SCPMG Genetics department perceived?**

- Almost all (91%) reported that it was “easy” or “very easy” for their patients to be seen by the Genetics Department.
- Nearly all (97%) reported that the Genetics department was “helpful” or “very helpful” in addressing their patients’ questions and concerns.
- Nearly every respondent (97%) indicated that the Genetics Department is “very valuable” or “valuable” to the organization.
- 98% believe that the Genetics Department staff are “expert” or “highly expert” in the area of genetics.

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### The New Los Angeles Times

Monday, November 1, 2010

**Kaiser Genetics: Wave of the Future**

Genetics department in Southern California integrated into standard medical processes.

*Primary care & subspecialties say medical genetics is “vital” to their practice.*

Family history is an essential tool in health promotion and disease prevention for all SCPMG members.

Genetics databases allow tracking of individuals and families with genetic disorders to provide state of the art care and measure outcomes of interventions.

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**Department of Health and Human Services Names Kaiser Genetics “Ideal U.S. Model”**

- Pharmacogenomic discoveries lead to expanded screening of new targeted populations.
- DNA based cancer screening enables pre-symptomatic interventions.
- Genetics provides a “family focus” for treatment of disease.
- Comprehensive, accurate family history data leads to more efficient use of healthcare resources.
The New Los Angeles Times
Tuesday, November 2, 2010
Kaiser Genetics In Southern California: Teams From Around The World Seek To Emulate Their Model
Studies Published By SPCM Genetics Department
Demonstrate Improved Health Outcomes

- Enhanced family communication of genetic risk factors improves awareness of carrier testing options
- Cultural competence in cancer genetic counseling results in improved cancer screening rates in high risk families across all ethnic groups.
- Clinician and patient genetics education projects lead to increased referral rates for patients with inherited risk factors
- Establishment of genetics peer review process promotes high reliability care and improved patient outcomes

Chinese delegation meets with SPCM Genetics Counselors Suzanne Poulson and Jeffrey Greenberg to discuss Kaiser’s innovative approaches to cultural competence in genetic counseling

The New Los Angeles Times
Wednesday, November 3, 2010
Kaiser Delivers “Personalized Medicine”: One Size Does Not Fit All

Our genetic make-up determines how we respond to medications.

Were she not a Kaiser Permanente member, Mrs. Johnson, a 73-year old resident of Hollywood, California, might not have received the most appropriate treatment for prevention of potentially lethal blood clots after breaking her hip from a fall at home.

However, since 2008, doctors at Kaiser Permanente Medical Centers have been using genetic testing to determine the appropriate dose for initiation of treatment with the blood thinner Coumadin. The test is simple and convenient. A small blood sample is taken and analyzed in the local laboratory. The result is available at “point of care” (at the patient’s bedside), allowing Kaiser Permanente physicians to make fast and accurate treatment decisions for patients like Mrs. Johnson.
A Mother’s Story
The Kaiser Genetics Department allowed me to enjoy my pregnancy without fear that my baby would have cystic fibrosis like her older brother. I was able to get testing in the first trimester and by the time I was in my second trimester I knew my baby would not be born with this terrible disease.

Kaiser Genetics: A Positive Influence on patient’s lives

Genetics made my practice more effective through education, genetic counseling and testing of high-risk patients and their families. They enabled me to prevent colorectal cancer in patients through surgical interventions in those identified as having high lifetime risks of the disorder.

Colorectal Surgeon’s Story

Thursday, November 4, 2010

The New Los Angeles Times

Frank and Ernest

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