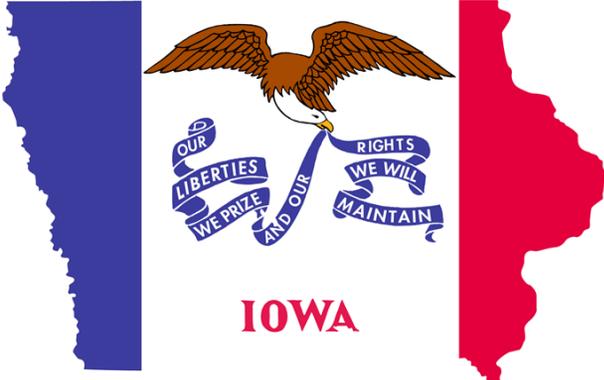


Regional Genetic Consultation Service

Contract 5885BD01
July 1, 2014 through June 30, 2015





Congenital & Inherited Disorders

Division of Health Promotion & Chronic Disease Prevention

Phone: 1-800-383-3826

www.idph.state.ia.us/genetics/default.asp



Regional Genetic Consultation Service:

a contract established by IDPH in the Division of Health Promotion & Chronic Disease Prevention, Contract # 5885BD01 Administrative Code, Chapter 4:641-4.5(136A)

Iowa Department of Public Health Advancing Health Through the Generations

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**In collaboration with the
Iowa Department of Public Health
&
The Stead Family Department of Pediatrics
Division of Medical Genetics
University of Iowa Children's Hospital
&
Clinical Outreach Services
University of Iowa Hospitals and Clinics**

What is the Regional Genetic Consultation Service?

Iowa Administrative Code 641—4.5(136A) Regional Genetic Consultation Service (RGCS). This program provides comprehensive genetic and genomic services statewide through outreach clinics.

4.5(1) Provision of comprehensive genetic and genomic services. The department shall contract with the Division of Medical Genetics within the Stead Family Department of Pediatrics at the University of Iowa to provide genetic and genomic health care and education outreach services for individuals and families within Iowa.

4.5(2) Clinical services. The services provided may include, but are not limited to: diagnostic evaluations, confirmatory testing, consultations by board-certified geneticists, genetic counseling, medical case management and referral to appropriate agencies.

Why Does the Regional Genetic Consultation Service Exist?

Purpose: to provide genetic health care services and education for individuals and families within the state of Iowa through statewide outreach services, in order to promote health, prevent disease, reduce the incidence of congenital disorders and improve outcomes for those with congenital defects and genetic disorders.

- Approximately 38,000 babies are born in Iowa every year. About 1500, or approximately 4%, are born with a congenital or inherited disorder.
- In addition, there are many chronic diseases, cancers, and mental disorders which are known to have a genetic component.
- With early diagnosis and medical treatment, complications from serious conditions, such as intellectual disabilities or even death, may be prevented, and disabilities may be eliminated or reduced.
- Genetic counseling and case management are an integral part of the genetics services provided, which helps to ensure the best possible outcome for patients and families.

The Regional Genetic Consultation Service assures that genetic specialists are available to Iowans to provide medical consultation and genetic counseling.

What is Genetic Counseling?



The purpose of genetic counseling is to provide information and support to individuals and families at risk for having, or who already have, a congenital defect or genetic disorder. Genetic counseling helps the individual or family:

- To comprehend the medical facts, including the diagnosis, probable disease course, and available treatment/management;
- To understand the way heredity contributes to the disease and the risk of recurrence for themselves and other family members;
- To understand the options available to deal with the risk of recurrence;
- To identify those beliefs, values, goals and relationships affected by the risk for or presence of a hereditary disease;
- To choose the course of action that seems most appropriate to them in view of their risk, their family goals, and their ethical and religious beliefs; and
- To make the best possible adjustment to the disorder or risk of occurrence of that disorder, or both, by providing supportive counseling and making referrals to appropriate specialists, social services, and family and patient support groups.

Thompson & Thompson, Genetics in Medicine, Edition 7

In the RGCS, genetic counseling services are provided by a team of medical specialists which includes Board Certified Medical Geneticists, Nurse Practitioners, Genetic Counselors and Genetic Nurse Specialists.

A Patient Story

Introduction: Noonan syndrome

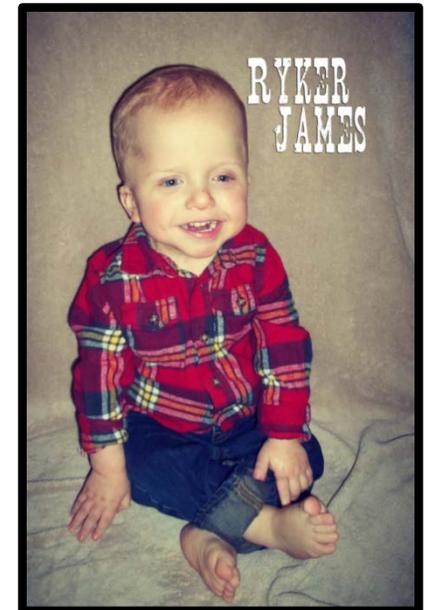
- A genetic disorder characterized by short stature, congenital heart defects, and developmental delay of variable degree.
- Cause: a change or mutation in any one of 13 different genes can cause Noonan syndrome. Approximately 25% of affected individuals do not have an identifiable mutation on the current genetic testing panels, thus indicating there are likely other mutations in genes which are not yet identified that cause Noonan syndrome.
- One of the most prevalent genetic disorders. Affects between 1:1000 to 1:2500 individuals. Mild expression is likely to be overlooked and underdiagnosed.
- Seen in all races and socioeconomic backgrounds.
- Autosomal dominant inheritance with 50-50 chance of passing the underlying mutation to a child. Anywhere from 30-75% of individuals are the first affected person in their family due to a new mutation in the responsible gene.

Features and Characteristics:

Associated symptoms and findings may vary greatly in range and severity, even within families. The same Noonan syndrome gene mutation present in different members of the same family (i.e. brothers, sisters, grandparents, parents and children) can result in Noonan syndrome with widely varying degrees of symptoms. However, common features include:

- Short stature
- Congenital heart defects
- Developmental delay of variable degrees
- Broad or webbed neck
- Unusual chest shape (called pectus carinatum or pectus excavatum)
- Undescended testes in males
- Characteristic facial features such as widely spaced and down-slanting eyes, low set and posteriorly rotated ears, vivid blue-green eyes, high arch in the roof of the mouth
- Buildup of fluid causing swelling, most commonly in the hands, feet, ankles, or legs (lymphedema)
- Bleeding problems (excessive bruising, nosebleeds, prolonged bleeding following surgery or injury)

Other features/complications may include: feeding issues in infancy, learning disabilities, kidney issues, hearing problems, cardiomyopathy, delayed puberty in males, eye abnormalities such as lazy eye, enlarged liver or spleen, and possible increased risk for cancer.



*Permission was obtained from Ryker's parents to include his photo in this report. Family supplied the photo.

Diagnosis:

The diagnosis of Noonan syndrome can often be made by a medical geneticist through physical exam and review of clinical history. Genetic testing, however, can be useful for prognosis and surveillance since mutations in certain genes which cause Noonan syndrome may put an individual at any increased chance for specific health concerns. As an example, patients with a mutation in the *RAF1* gene have a 95% chance to develop hypertrophic cardiomyopathy (a thickening of the heart muscle that forces the heart to work harder to pump blood). This is compared to the 20-30% risk of cardiomyopathy for individuals with mutations in other genes that cause Noonan syndrome.

Treatment:

There is no cure for Noonan syndrome, but many of the symptoms/complications can be treated.

What to Expect:

Given the associated problems, multiple medical specialist visits may be required throughout life. Early intervention services are important to ensure that affected individuals reach their potential.

Personal Stories:

RGCS sees many Iowa patients with birth defects and inherited disorders like Noonan syndrome. Through correct diagnosis, education and care coordination, these children and adults can be helped to lead healthy and productive lives. Many of the patients seen in the genetics clinics through the Regional Genetic Consultation Service and University of Iowa are similar to the stories shown on the website:

<http://www.childrenwithspecialneeds.com/>

Common Types of Genetic & Congenital Disorders

Introduction to Genetic and Congenital Disorders:

A genetic disorder is a disease caused by an abnormality in an individual's DNA (deoxyribonucleic acid). DNA is the code that provides the instructions or blueprint that tells each cell in our body what it needs to know to grow and develop properly. DNA instructions are organized into segments called genes. Genes are packaged in compact units called chromosomes. Humans have 46 chromosomes and approximately 20,000 genes. Abnormalities in DNA can range from a small mutation or change in the DNA code of a single gene to the addition or subtraction of an entire chromosome or segment of a chromosome. Some genetic disorders are inherited and some are new, occurring for the first time in an individual.

Not all congenital disorders are caused by a genetic abnormality. In some cases these disorders are caused by environmental exposures during pregnancy. These exposures could include:

- infectious agents (i.e. rubella, herpes and toxoplasmosis),
- physical agents (i.e. high levels of radiation, high fevers and uterine abnormalities),
- drugs and chemicals (i.e. prescription medications, recreational drugs, alcohol, tobacco and toxic chemicals) and;
- maternal factors (i.e. poorly controlled diabetes or PKU and malnutrition).

Still other congenital disorders, like cleft lip and palate or spina bifida, are multifactorial in nature, caused by a combination of environmental factors and one or more genes. For more information: <http://ghr.nlm.nih.gov/>.

Table 1

Examples of Genetic/Congenital Disorders	
Angelman Syndrome	Lesch-Nyhan Syndrome
Apert Syndrome	Lissencephaly
Autism Spectrum Disorders	Marfan Syndrome
CHARGE Syndrome	Mitochondrial Disorders
Cleft lip and / or Palate	Neurofibromatosis
Cornelia de Lange Syndrome	Noonan Syndrome
Cri Du Chat Syndrome	Phenylketonuria (PKU)
Dandy-Walker Syndrome	Prader-Willi Syndrome
Deletion Syndromes	Rett Syndrome
Developmental / Growth Delay	Rubinstein-Taybi Syndrome
DiGeorge / Velocardiofacial Syndrome	Sanfilippo Syndrome
Down Syndrome	Septo-Optic Dysplasia
Familial Cancer Syndromes	Smith-Lemli-Opitz Syndrome
Fetal Alcohol Syndrome	Smith-Magenis Syndrome
Fragile X Syndrome	Spina Bifida
Ehlers Danlos Syndrome	Sturge-Weber Syndrome
Huntington's Disease	Trisomy 13 or 18
Hydrocephalus	Tuberous Sclerosis
Kabuki Syndrome	Turner Syndrome
Klinefelter Syndrome	Williams Syndrome
Laurence-Moon-Bardet-Biedl Syndrome	Von Hippel-Lindau Syndrome

Often there is no cure, but treatment and medicines can help manage some of the symptoms.

Table 2

Incidence / Prevalence of Genetic & Congenital Disorders in Iowa		
<i>Genetic/Congenital Disorders</i>	<i>Incidence Of Genetic / Congenital Disorders</i>	<i>Estimated # Newborns affected</i>
Congenital Malformations (Newborns)	~4%	1,500/yr
• Chromosome Abnormalities	~1%	400/yr
• Multifactorial/ Single gene / Environmental	~3%	1,100/yr
<i>Other Diseases/disorders with Genetic components</i>	<i>Incidence or Prevalence</i>	<i>Estimated # Affected</i>
Cancers – 5-10% have a strong inherited susceptibility	17,500 Iowans will be diagnosed with cancer/yr	875-1,750 new cancers/yr with genetic component
Chronic Diseases (heart disease, diabetes) - ~10% have a significant genetic component	>1 million Iowans suffer from at least 1 chronic disease	>100,000 individuals with these diseases have a genetic component to their disease
Intellectual Disability (or ID, formerly referred to as Mental Retardation or MR) ~ 50% of ID has a significant genetic component	~24,000 Iowans are estimated to have a diagnosis of ID	~12,000 of those with ID would be expected to have a genetic component to their condition

*More than 20% of infant deaths are caused by birth defects or genetic conditions (e.g. congenital heart defects, abnormalities of the nervous system, or chromosomal abnormalities).

**Approximately 10% of all adult hospitalizations and 30% of all childhood hospitalizations are due to a genetically related problem.

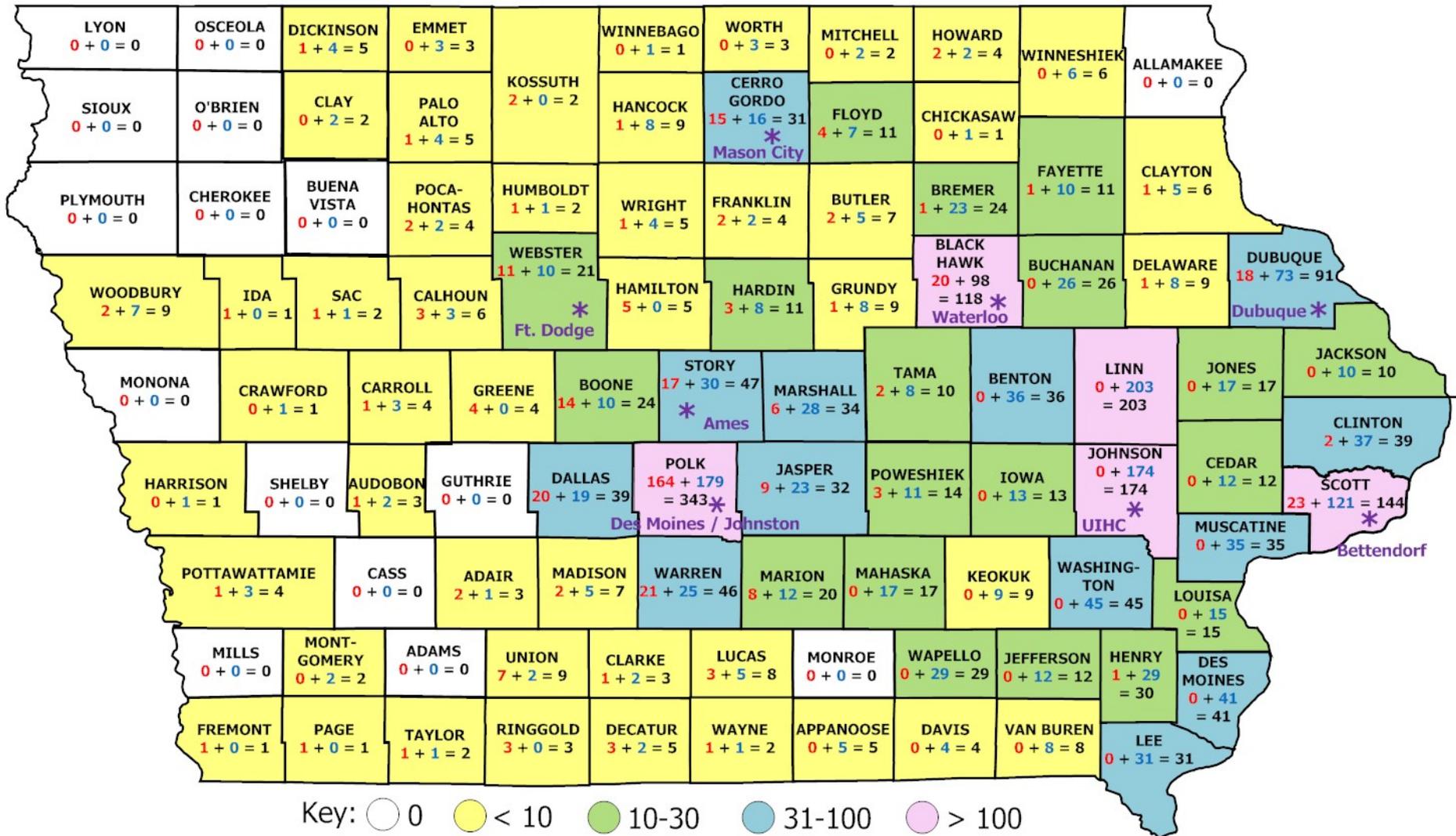
References:

- <http://www.kumc.edu/gec/prof/prevalnc.html>
- <http://www.namiiowa.com/>
- <http://www.netwellness.org/healthtopics/idbd/2.cfm>
- <http://www.google.com/publicdata?ds=uspopulation&met=population&idim=state:19000&q=iowa+population>
- <http://www.fightchronicdisease.org/>
- <http://www.cancer-rates.info/ia>
- <http://www.familiesfightingautism.org>

Figure 1

FY 2015 RGCS & UIHC Genetics Patient Census by County

(RGCS is shown in red; UIHC is shown in blue)



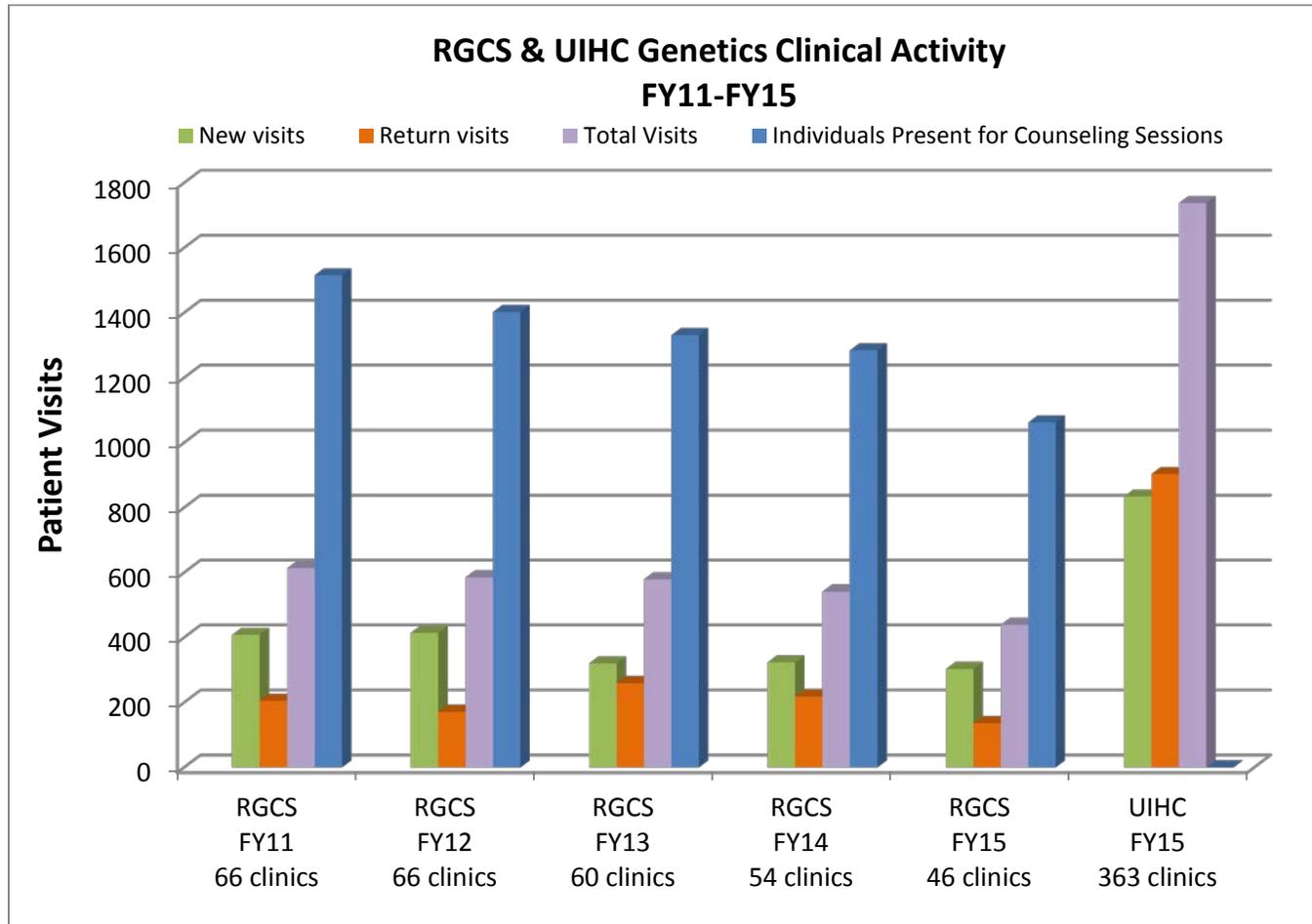
Key: ○ 0 ● < 10 ● 10-30 ● 31-100 ● > 100

Total Patients = 2176; * Clinic Sites

Outliers: Patients = 74 (included in total) Illinois, Nebraska, North Carolina, South Dakota & Wisconsin

RGCS & UIHC Clinical Activity

Figure 2



*During the spring of FY15, two genetic nurse/counselors retired, thus clinic numbers were slightly decreased to allow for adequate transition as two new genetic counselors were not hired until May and June 2015.

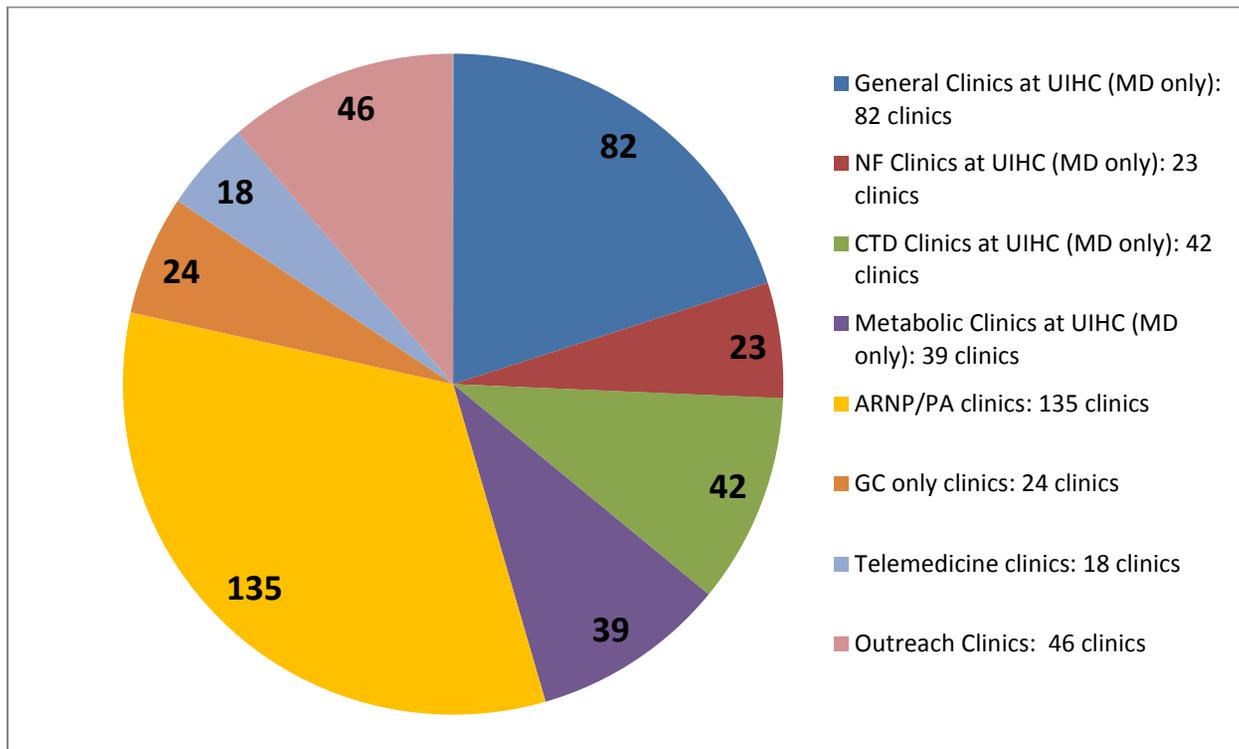
Table 3

Patient Visits	RGCS FY11 66 clinics	RGCS FY12 66 clinics	RGCS FY13 60 clinics	RGCS FY14 54 clinics	RGCS FY15 46 clinics	RGCS & UIHC* FY15 409 clinics
New visits	408	414	320	323	303	1137
Return visits	205	171	259	218	136	1039
Total Visits	613	585	579	541	439	2176
Individuals Present for Counseling Sessions	1515	1402	1331	1284	1062	NA

*New for FY15: all UIHC clinic sites are now included in the numbers

Total RGCS & UIHC Clinics (MD, ARNP, GC only) FY15

Figure 3



RGCS Patient Population by Age and Gender

Table 4

AGE	FY 2011		FY 2012		FY 2013		FY 2014		FY 2015		% Gender by Age (5 year average)		
	Female	Male	Female	Male									
1-364 days	29	28	43	34	28	29	32	29	28	26	5.79%	5.28%	
1-4 yrs	76	96	83	58	78	61	60	72	39	60	12.16%	12.55%	
5 YEAR TOTAL (UNDER 5 YEARS of AGE)												36%	
5-9 yrs	59	80	59	57	59	60	51	67	33	54	9.44%	11.51%	
10-14 yrs	36	44	45	51	43	57	50	55	24	42	7.16%	9.01%	
15-19 yrs	30	30	30	28	27	29	24	24	27	25	4.99%	4.92%	
5 YEAR TOTAL (5-19 YEARS of AGE)												47%	
20-29 yrs	25	12	23	14	29	10	19	11	16	12	4.05%	2.13%	
30-39 yrs	30	10	23	16	25	16	17	14	18	6	4.09%	2.24%	
40-49 yrs	10	4	11	7	10	5	5	2	12	3	1.74%	0.76%	
50-59 yrs	6	1	6	1	3	6	6	2	6	0	0.98%	0.36%	
60-69 yrs	2	0	3	1	3	1	1	0	4	4	0.47%	0.22%	
70 & older	1	0	1	1	0	1	0	0	0	0	0.07%	0.07%	
5 YEAR TOTAL (OVER 20 YEARS of AGE)												17%	
Totals	304	305	327	268	305	275	265	276	207	232	50.94%	49.06%	100%

RGCS & UIHC Total Patient Population by Age and Gender (FY 2015)

Table 5

AGE	UIHC & RGCS FY 2015		% Gender by Age (1 year average)		
	Female	Male	Female	Male	
1-364 days	141	161	6.47%	7.40%	
1-4 yrs	196	216	9.01%	9.93%	
1 YEAR TOTAL (UNDER 5 YEARS of AGE)					33%
5-9 yrs	166	220	7.63%	10.11%	
10-14 yrs	147	157	6.76%	7.22%	
15-19 yrs	129	99	5.93%	4.55%	
1 YEAR TOTAL (5-19 YEARS of AGE)					42%
20-29 yrs	114	68	5.24%	3.13%	
30-39 yrs	124	31	5.70%	1.42%	
40-49 yrs	80	29	3.68%	1.33%	
50-59 yrs	48	14	2.21%	0.64%	
60-69 yrs	17	14	0.78%	0.64%	
70 & older	4	1	0.18%	0.05%	
1 YEAR TOTAL (OVER 20 YEARS of AGE)					25%
Totals	1166	1010	53.59%	46.41%	100%

Services Provided by the Regional Genetic Consultation Service Per Contract

The RGCS contract for FY 2015 required a minimum of 135 clinics be held via on site services or telehealth. Expectations and services provided for individuals and families who have, are suspected to have, or are at risk for a congenital and/or inherited disorders included:

Physician Evaluation and Medical Management

- ◆ **Consultation by board-certified geneticists**
- ◆ **Diagnostic Evaluation**
 - detailed physical examinations
 - review of family history and medical records
 - diagnostic and confirmatory testing
- ◆ **Management of health care concerns**
 - medical management to slow progression of disease, control pain and treat symptoms
 - ongoing monitoring/ follow-up
 - referrals to other specialists as needed

Genetic Counseling, Care Coordination, Education and Advocacy

- ◆ **Patient and Family Education**
 - written and verbal information provided specific to diagnosis, disease process, treatment & management
 - genetic counseling for patient and families including information regarding recurrence risks and options available to deal with the recurrence risk
 - anticipatory guidance regarding prognosis and level of disability
 - information about guardianship, advance directives & living wills, as appropriate
 - education of family, school personnel, employers, childcare providers and others
 - updates on research for patients, families and healthcare providers
- ◆ **Patient & Family Support Services**
 - phone triage and assistance with daily management of emotional, social and physical aspects of the disorder.
 - advocacy in communicating with educators, employers, health insurers and others.
 - assistance in identifying social & financial services
 - referrals to educational resources, home health care, respite and hospice services
 - referrals to patient and family support groups
 - access to research opportunities, as desired

Medical Geneticist Consultation to Iowa Physicians available 24 hours per day, 365 days per year.

Educational presentations and activities

- provide educational presentations for medical personnel, patient/family groups, support groups, graduate and undergraduate students
- provide educational opportunities through observational and mentoring activities for medical and nursing students and other ancillary professionals
- participate in regional collaborative groups

In addition, the RGCS staff:

- participates in the activities of the Center for Congenital and Inherited Disorders (CIDAC) Advisory Committee, providing assistance and technical support to the IDPH.
- coordinates and integrates services with other programs serving similar purposes and populations.

Evaluation of RGCS FY 2015 Has RGCS Met Contract Objectives?

Meeting Contract Objectives:

- In FY 2015, the RGCS program (with the new inclusion of University of Iowa clinic sites) conducted 409 clinics in 8 Iowa communities, providing 2176 clinic visits for patients and/or families (Figures 1, 2, & 3; Tables 3, 4 & 5). Of these, 46 clinics were held at regional outreach sites, 345 clinics were held at the University of Iowa Hospitals and Clinics, and 18 clinics were held via telemedicine with the provider located at the University of Iowa Hospitals and Clinics. At the regional outreach locations, approximately 1,062 patients, family members and caregivers participated in these visits. The clinics were staffed by geneticists, nurse practitioners, physician assistants, and genetic/nurse counselors who provided diagnostic evaluation and medical management, genetic counseling, care coordination, education, support, advocacy and follow-up.
- Patients of all ages are seen in the RGCS and UIHC clinics (Tables 4 & 5). The majority of the regional outreach clinics (33 of the 46) were held in the Des Moines/Johnston and Ames sites, with another 4 being held in Fort Dodge and Mason City, in order to help increase accessibility for individuals living on the western side of the state. Figure 1 displays the county of residence for individuals seen in clinic.
- The RGCS staff is actively involved in education of a variety of health care professionals and students on a regular basis. Many hours are spent with genetics and pathology fellows; medical students; graduate nursing students; residents from family practice, pediatrics, pathology and dentistry; genetic counseling interns and cytogenetics staff, both in the clinical and the academic setting. Many genetics staff facilitate the genetics small group sessions for Mechanisms of Health and Disease for the 1st year medical students at the University of Iowa. Genetics staff also take turns presenting current/interesting journal articles in the Genetics Journal Club which meets weekly for interested University staff and students.
- RGCS staff is also involved in numerous presentations on a variety of genetics related topics. Presentations have been made this past year to physician groups, residents and medical students, nurses, and families. The genetics staff organizes 2 symposiums yearly: one for patients/families with Neurofibromatosis and another for patients/families with metabolic conditions requiring treatment with low protein foods. Members of the Regional Genetic Consultation Service team have also done poster

presentations at national genetics meetings including: the American College of Medical Genetics and the American Society of Human Genetics.

- RGCS staff participate in the activities/work groups of the Heartland Regional Genetics and Newborn Screening Collaborative.
- RGCS staff continue to participate in CIDAC meetings and activities.
- RGCS staff collaborates with Child Health Specialty Clinics, Early ACCESS and the statewide Area Education Agencies, Newborn Screening and other programs to coordinate and integrate services with other programs serving similar purposes and populations.

Since April 1, 2012, the oversight of the organization and structure of the RGCS program has been provided by the Clinical Outreach Service of the University of Iowa Hospitals and Clinics. The UIHC Outreach Service provides the following services to the RGCS program: clinic scheduling; clinic site negotiations, contract development and payment of site rental fees; transportation to and from clinic for clinic staff, medical personnel for onsite clinic registration; salary support for physicians' and nurse practitioners' for the time spent traveling to and from clinic and the hours of the clinic; and distribution of clinic summary letters. The money received from fee for service for patient visits is used by UIHC Clinical Outreach Service to pay for the above services.

The money that is received from the Iowa Department of Public Health contract supports: the salaries of the genetic/nurse counselors in UIHC Genetics program who provide genetic counseling and case management/care coordination services for the patients and families served in the genetics program; the salaries of the Division of Medical Genetics administrative/ support staff that assist with the duties of the RGCS program; and the purchase of supplies needed to carry out the clinics and provide patient/family care including patient education materials.

National Performance Measures / Priorities Outlined in Iowa's Family Health Plan that RGCS Participated in During FY 2015

Assure families of children with special health care needs ages 0-18 years partner in decision making at all levels and are satisfied with the services they receive.

National Measure #2

A central part of the services provided by the RGCS staff is the education and counseling of families/patients regarding their specific disease process, recurrence risks in future pregnancies, recommended treatment/management and current research with an ultimate goal of empowering them to make choices regarding their health care that best meets their individual/family needs. Referrals to support groups, the Parent Consultant Network through Child Health Specialty Clinics and other medical resources also help to assure family involvement/decision making in meeting the child's health care needs. To assess satisfaction with genetics services, families are provided with an opportunity to complete a satisfaction survey following their clinic visit. This information is used to improve services whenever possible.

Assure children with special health care needs age 0 to 18 receive coordinated, on-going, comprehensive care within a medical home.

National Measure #3

Many of the patients seen in RGCS and UIHC clinics have very complex health care needs. The genetics staff strives to provide a comprehensive assessment of the individual's medical, social and educational needs. Although only a small number of patients actually have an designated medical home, clinic summary and results letters are used to communicate the genetics assessment, recommendations/plan of care and results of evaluations to the primary care physician/medical home/referring physician to assist with coordination of services. Whenever possible/available, local resources are utilized to meet the patient's special health/social/educational/emotional needs. Follow-up appointments in genetics allow for: re-assessment and evaluation of patient's needs; review of appropriateness of current management; further education of patient, family and caregivers; and identification of need for new services in order to assure on-going comprehensive care.

Assure families of children with special health care needs age 0 to 18 have adequate private and/or public health insurance to pay for the services they need.

National Measure #4

As part of their role with families and patients, the RGCS staff assists families/parents to identify social services and financial resources to meet their health care needs, aiding in the application process when needed. Staff members serve as advocates for patients by communicating with health insurers regarding the importance of appropriate diagnostic testing; appropriate screening, management and treatment for specific (often rare) genetic disorders and by providing documentation and educational resources as needed. RGCS staff also participate in the Center for Congenital and Inherited Disorders Advisory Committee where there is an opportunity to have dialogue with members of the Iowa Insurance Commission regarding health care needs of children/patients with complex health conditions.

Assure families of children with special health care needs ages 0 to 18 have access to community-based services that are organized for easy use.

National Measure #5

The RGCS program continues to provide outreach genetics services throughout eastern and central Iowa in an attempt to improve access of these unique services for Iowans. Clinic sites utilized are generally health care facilities that are easily accessible and in close proximity to a variety of other health care services. As previously mentioned, whenever possible/available, local/community resources are utilized for diagnostic and management/treatment services to meet the patient's needs.

Assist youth with special health care needs to receive services necessary for successful transition to all aspects of adult life, including adult health care, work, and independence.

National Measure #6

The RGCS staff provide care to individuals with complex health and developmental needs of all ages. Because of this, they are acutely aware of the need to assist families to transition these youth to adult programs and services as they reach adulthood. The RGCS staff assist with education of families regarding guardianship issues; serve as advocates for patients and families to assist with identification of local resources for adults; and make referrals for medical, social and educational services as needed.

Barriers, Challenges and Additional Facts

The RGCS program has held 2,762 regional outreach clinics with more than 27,212 patient/family visits over the past 39 years. In addition, 363 clinics were also held at UIHC and via telemedicine services in FY15. As the public's knowledge and awareness of genetic services continues to expand, the referrals for genetic evaluations and services (for RGCS and UIHC clinics) continue to grow, exceeding the clinic and staff capacity. In September of 2013, a new RGCS/ UI genetics and cytogenetics physician joined the staff. In May 2015, two genetic nurse/counselors retired and clinic visits were decreased in the 6 weeks leading up to their retirement to allow for adequate patient care coordination, completion of follow-up, and transition. Two new genetic counselors were subsequently hired in May 2015 and June 2015. Finding adequate numbers of qualified genetics personnel (physicians and counselors/nurses) continues to be a significant challenge. Recruitment for physicians and genetic nurse/counselors is ongoing.

The typical wait time for a genetics appointment for a “non-critical” patient is often 6-8 months. As a result, the backlog of patients awaiting appointments, especially those awaiting follow-up, continues to be a major issue of concern. The genetics staff continues to deal with this by: timely review and assessment of patient records to assess acuity; prioritizing patient's/physician's requests for clinic appointments; increasing the number of patient appointments per clinic day; overbooking appointments at the University on non-clinic days when needed; and providing medical recommendations for local providers to begin the evaluation process prior to the genetics appointment.

Transportation also continues to be a significant barrier for some families, particularly for those with limited resources who live a distance from the clinic sites. Resources, such as TMS (Non-Emergency Medical Transportation Service), have often been sorely inadequate and frequently obstructive for families. Because there are currently no RGCS clinics on the western part of the state, families from that area are sometimes referred to resources in surrounding states that are closer to their home. When necessary and possible, we assist families with identification of local transportation resources, (i.e. support group members) to help transport them to their appointments.

The RGCS staff has been working with staff from Child Health Specialty Clinics, Clinical Outreach Services and the Stead Family Department of Pediatrics to explore the feasibility of expanding telemedicine/E-medicine genetics clinics in order to increase access for Iowa citizens. The logistics of establishing these clinics has proven to be quite challenging. Funding for equipment and staff, staffing at the site of the telemedicine clinic for patient registration and vitals, scheduling of patients/equipment and site, and 2nd party payer reimbursement are a few of the areas that have served as barriers to the establishment to these clinics. A few appointments have been attempted with patients seen by two of the genetics ARNPs with variable success. We will continue to work to see if this can become a viable part of the genetic services available to Iowans.

Insurance issues continue to be a major challenge for staff and families. This is an area that is constantly changing. It is extremely difficult for staff and families to understand and be able to verify if appointments and testing are a covered service in a particular health insurance plan. Many procedures such as MRIs and echocardiograms require prior approval. Preauthorization often requires significant staff time. Staff completes preauthorization paperwork including documentation of the reason for the evaluations and how it will impact the patient's care. At times, phone conversations between the genetics physician provider and the insurance medical director are required to get approval. Although in some cases, genetic testing may not require prior approval, but there is no assurance for the family that this testing will be a covered service. Since genetic laboratory testing is often expensive, beyond what the typical family can afford, they are often wary of proceeding with recommended testing. Dealing with insurance issues is not only time consuming, but can significantly affect our ability to provide appropriate genetic health care and carry out recommended evaluations/testing for the patients and families we serve.

Regional Genetic Consultation Service Director Statements

Val C. Sheffield, MD, PhD
Professor of Pediatrics
Medical Director, Division of Medical Genetics
Howard Hughes Medical Institute Investigator

Pamela Trapane, MD
Clinical Associate Professor of Pediatrics
Clinical Director, Division of Medical Genetics

Medical genetics started at the beginning of the 20th century when it was recognized that the occurrence of some human diseases in families could be explained by Mendel's principles. During the past several decades, Medical genetics has grown into a medical specialty concerned with the diagnosis and management of many disorders, including both rare and common diseases. It is now recognized that nearly all diseases have a genetic component. Medical genetics focuses not solely on individual patients, but on the entire family.

The obtaining of a comprehensive family history is an important first step in the practice of good medicine as it can lead to diagnosis, prognosis, and assessment of recurrence risk in family members. This information, in turn, plays a key role in the proper counseling, management and disease prevention for patients and their families. Regional Genetic Consultation Service began nearly forty years ago with the goal of providing Iowa patients and families with proper medical treatment and information utilizing the most current genetic information and resources.

In the past, the Human Genome Project determined the complete sequence of the human genome. This has facilitated the increasingly rapid identification of genes and genetic variation associated human disease. Along with other advances including information management and biological technologies, the practice of medical genetics is revolutionizing medicine by improving diagnosis, disease

prevention and treatment based on genetic information. The opportunities afforded by the Human Genome Project and other advances in genetics are being and will be utilized by the Regional Genetic Consultation Service program for the betterment of the health of Iowans in a number of ways including the following:

Diagnosis:

Individuals with birth defects are undergoing high-resolution genome testing to identify genetic duplications and deletions causing congenital malformations and other disorders. The resolution of such testing using DNA microarray technology (DNA chips) makes it possible to identify smaller and smaller abnormalities.

The identification of human genes and the mutations associated with hundreds of genetic diseases are making it possible to make disease diagnoses based on genetic sequences, thus differentiating between highly heritable diseases and diseases with low heritability. Newer laboratory methods offer the ability to rapidly and accurately sequence panels of genes associated with specific genetic conditions. In addition, whole exome sequencing is an emerging technology that will be of benefit to patients with rare undiagnosed disorders.

Disease Prevention:

Genetic testing is making it possible to detect individuals at risk for disease even prior to onset of symptoms. In some cases, interventions are available to lower the risk of developing the disease, or at a minimum lead to early identification and management of disease symptoms.

Treatment:

In some cases, genetic analysis allows for selection of the most effective treatment. For example, genetic analysis of a tumor sample from a cancer patient can aid in the selection of the most effective chemotherapy for the patient, as well as avoiding adverse reactions to a chemotherapeutic agent in a particular patient.

While the above examples are currently possible for some disorders, the future holds the extension of diagnostic, prevention and treatment modalities to countless additional diseases and individuals. The need for genetic services continues to expand given the ongoing and rapid development of more sensitive and affordable genetic tests and novel therapies for disease treatment. The Regional Genetic Consultation Service program will continue to be key component to the delivery of genetic services to patients and families within the State of Iowa.