Regional Genetic Consultation Service

Contract 5887BD01
July 1, 2016 through June 30, 2017
Congenital & Inherited Disorders
Division of Health Promotion & Chronic Disease Prevention
Phone: 1-800-383-3826
http://idph.iowa.gov/genetics

In collaboration with the
Iowa Department of Public Health
&
The Stead Family Department of Pediatrics
Division of Medical Genetics
University of Iowa Stead Family 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 cognitive 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, 8th Edition*

In the RGCS, genetic counseling services are provided by a team of medical specialists, which includes Board Certified Medical Geneticists, Nurse Practitioners, Physician Assistants, Genetic Counselors, and Nurse Clinicians.
**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 15 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, which affects between 1:1000 to 1:2500 individuals. Mild forms of the disorder are 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. 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 an increased chance for specific health concerns. As an example, patients with a mutation in the \textit{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/featured-kids/

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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 blueprints 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)
- maternal factors (i.e. poorly controlled diabetes or PKU and malnutrition).

Still other congenital disorders, such as 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/](http://ghr.nlm.nih.gov/).

<table>
<thead>
<tr>
<th>Examples of Genetic/Congenital Disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Angelman syndrome</td>
</tr>
<tr>
<td>Apert syndrome</td>
</tr>
<tr>
<td>Beckwith-Wiedemann syndrome</td>
</tr>
<tr>
<td>CHARGE syndrome</td>
</tr>
<tr>
<td>Cleft lip and / or Palate</td>
</tr>
<tr>
<td>Cornelia de Lange syndrome</td>
</tr>
<tr>
<td>Developmental / Growth Delay</td>
</tr>
<tr>
<td>Down syndrome</td>
</tr>
<tr>
<td>Dysmorphic facial characteristics</td>
</tr>
<tr>
<td>Ehlers Danlos syndromes</td>
</tr>
<tr>
<td>Fabry Disease</td>
</tr>
<tr>
<td>Familial Cancer Syndromes</td>
</tr>
<tr>
<td>Fetal Alcohol Syndrome</td>
</tr>
<tr>
<td>Fragile X syndrome</td>
</tr>
<tr>
<td>Galactosemia</td>
</tr>
<tr>
<td>Hereditary Hemorrhagic Telangiectasia</td>
</tr>
<tr>
<td>Hunter syndrome</td>
</tr>
<tr>
<td>Hydrocephalus</td>
</tr>
<tr>
<td>Kabuki syndrome</td>
</tr>
<tr>
<td>Klinefelter syndrome</td>
</tr>
<tr>
<td>Leigh syndrome</td>
</tr>
</tbody>
</table>

Often there is no cure, but treatment and medicines can help manage some of the symptoms.
Table 2

<table>
<thead>
<tr>
<th>Genetic/Congenital Disorders</th>
<th>Incidence Of Genetic / Congenital Disorders</th>
<th>Estimated # Newborns affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital Malformations (Newborns)</td>
<td>~4%</td>
<td>1,500/yr</td>
</tr>
<tr>
<td>• Chromosome Abnormalities</td>
<td>~1%</td>
<td>400/yr</td>
</tr>
<tr>
<td>• Multifactorial/ Single gene / Environmental</td>
<td>~3%</td>
<td>1,100/yr</td>
</tr>
</tbody>
</table>

Other Diseases/disorders with Genetic components

<table>
<thead>
<tr>
<th>Incidence or Prevalence</th>
<th>Estimated # Affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cancers – 5-10% have a strong inherited susceptibility</td>
<td>17,500 Iowans will be diagnosed with cancer/yr</td>
</tr>
<tr>
<td>Chronic Diseases (heart disease, diabetes) - ~10% have a significant genetic component</td>
<td>&gt;1 million Iowans suffer from at least 1 chronic disease</td>
</tr>
<tr>
<td>Intellectual Disability (or ID, formerly referred to as Mental Retardation or MR) ~ 50% of ID has a significant genetic component</td>
<td>~24,000 Iowans are estimated to have a diagnosis of ID</td>
</tr>
</tbody>
</table>

*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
FY 2017 RGCS & UIHC Genetics Patient Census by County
(RGCS is shown in red; UIHC is shown in blue)

Figure 1

Total Patients = 2749; * Clinic Sites

Outliers: Total Patients = 137 (included in total) - RGCS - 3; UIHC - 134

Outlier States Include: IL, KS, MN, MO, NE, TX, WI
*Prior to FY15, only patients seen in RGCS clinics were included in the annual reports. As of FY15, due to state contractual changes, all patients, including those seen in the RGCS clinics as well as UIHC clinic sites are included in the numbers.

**During FY17, RGCS clinic numbers were slightly decreased due to turnover of genetics physicians and genetic counseling staff maternity leave.
### Table 3

<table>
<thead>
<tr>
<th>Patient Visits</th>
<th>RGCS FY13 60 clinics</th>
<th>RGCS FY14 54 clinics</th>
<th>RGCS &amp; UIHC* FY15 409 clinics</th>
<th>RGCS &amp; UIHC* FY16 467 clinics</th>
<th>RGCS &amp; UIHC* FY17 514 clinics</th>
</tr>
</thead>
<tbody>
<tr>
<td>New visits</td>
<td>320</td>
<td>323</td>
<td>1137</td>
<td>1096</td>
<td>1590</td>
</tr>
<tr>
<td>Return visits</td>
<td>259</td>
<td>218</td>
<td>1039</td>
<td>1279</td>
<td>1159</td>
</tr>
<tr>
<td>Total Visits</td>
<td>579</td>
<td>541</td>
<td>2176</td>
<td>2375</td>
<td>2749</td>
</tr>
<tr>
<td>Individuals Present for Counseling Sessions</td>
<td>1331</td>
<td>1284</td>
<td>NA</td>
<td>NA</td>
<td>6321</td>
</tr>
</tbody>
</table>

NA: This statistic was not tracked during FY15 and FY16 for UIHC clinics and is thus not reportable for those years.

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

**It should be noted that others involved in the counseling sessions most often include parents, grandparents and siblings of patients. These individuals have much to gain from the understanding of the inheritance, interventions and natural history of the disorder found in their family.

### Total RGCS & UIHC Clinics (MD, PA, ARNP, GC only) FY17

*The above clinic counts are representative of regularly scheduled clinics. These do not take into account urgent add-on patients, which are often seen on non-clinic days.
## RGCS Patient Population by Age and Gender

**Table 4**

<table>
<thead>
<tr>
<th>AGE</th>
<th>FY 2013</th>
<th>FY 2014</th>
<th>FY 2015</th>
<th>FY 2016</th>
<th>FY 2017</th>
<th>% Gender by Age (5 year average)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1-364 dys</td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
</tr>
<tr>
<td>1-4 yrs</td>
<td>28</td>
<td>29</td>
<td>32</td>
<td>29</td>
<td>28</td>
<td>26</td>
</tr>
<tr>
<td>5-9 yrs</td>
<td>59</td>
<td>60</td>
<td>51</td>
<td>67</td>
<td>33</td>
<td>54</td>
</tr>
<tr>
<td>10-14 yrs</td>
<td>43</td>
<td>57</td>
<td>50</td>
<td>55</td>
<td>24</td>
<td>42</td>
</tr>
<tr>
<td>15-19 yrs</td>
<td>27</td>
<td>29</td>
<td>24</td>
<td>24</td>
<td>27</td>
<td>25</td>
</tr>
<tr>
<td>20-29 yrs</td>
<td>29</td>
<td>10</td>
<td>19</td>
<td>11</td>
<td>16</td>
<td>12</td>
</tr>
<tr>
<td>30-39 yrs</td>
<td>25</td>
<td>16</td>
<td>17</td>
<td>14</td>
<td>18</td>
<td>6</td>
</tr>
<tr>
<td>40-49 yrs</td>
<td>10</td>
<td>5</td>
<td>5</td>
<td>2</td>
<td>12</td>
<td>3</td>
</tr>
<tr>
<td>50-59 yrs</td>
<td>3</td>
<td>6</td>
<td>6</td>
<td>2</td>
<td>6</td>
<td>0</td>
</tr>
<tr>
<td>60-69 yrs</td>
<td>3</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>70 &amp; older</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td><strong>Totals</strong></td>
<td>305</td>
<td>275</td>
<td>265</td>
<td>276</td>
<td>207</td>
<td>232</td>
</tr>
</tbody>
</table>

### 5 YEAR TOTAL UNDER 5 YEARS of AGE

- **37%**

### 5 YEAR TOTAL 5-19 YEARS of AGE

- **48%**

### 5 YEAR TOTAL OVER 20 YEARS of AGE

- **15%**
## RGCS & UIHC Total Patient Population by Age and Gender (FY15 – FY17)

### Table 5

<table>
<thead>
<tr>
<th>AGE</th>
<th>FY 2015</th>
<th>FY 2016</th>
<th>FY 2017</th>
<th>% Gender by Age (5 year average)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Female</td>
<td>Male</td>
<td>Female</td>
<td>Male</td>
</tr>
<tr>
<td>1-364 dys</td>
<td>141</td>
<td>161</td>
<td>114</td>
<td>190</td>
</tr>
<tr>
<td>1-4 yrs</td>
<td>196</td>
<td>216</td>
<td>196</td>
<td>252</td>
</tr>
<tr>
<td><strong>5 YEAR TOTAL UNDER 5 YEARS OF AGE</strong></td>
<td><strong>31%</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>5-9 yrs</td>
<td>166</td>
<td>220</td>
<td>203</td>
<td>236</td>
</tr>
<tr>
<td>10-14 yrs</td>
<td>147</td>
<td>157</td>
<td>189</td>
<td>189</td>
</tr>
<tr>
<td>15-19 yrs</td>
<td>129</td>
<td>99</td>
<td>146</td>
<td>112</td>
</tr>
<tr>
<td><strong>5 YEAR TOTAL 5-19 YEARS OF AGE</strong></td>
<td><strong>44%</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>20-29 yrs</td>
<td>114</td>
<td>68</td>
<td>131</td>
<td>69</td>
</tr>
<tr>
<td>30-39 yrs</td>
<td>124</td>
<td>31</td>
<td>113</td>
<td>38</td>
</tr>
<tr>
<td>40-49 yrs</td>
<td>80</td>
<td>29</td>
<td>72</td>
<td>28</td>
</tr>
<tr>
<td>50-59 yrs</td>
<td>48</td>
<td>14</td>
<td>53</td>
<td>15</td>
</tr>
<tr>
<td>60-69 yrs</td>
<td>17</td>
<td>14</td>
<td>24</td>
<td>7</td>
</tr>
<tr>
<td>70 &amp;older</td>
<td>4</td>
<td>1</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td><strong>5 YEAR TOTAL OVER 20 YEARS OF AGE</strong></td>
<td><strong>25%</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Totals</td>
<td>1166</td>
<td>1010</td>
<td>1242</td>
<td>1136</td>
</tr>
</tbody>
</table>
Services Provided by the Regional Genetic Consultation Service Per Contract

The RGCS contract for FY 2017 required a minimum of 1,000 patient visits, including visits for counseling services, to 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 is 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

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**Evaluation of RGCS FY 2017**

**RGCS Met Contract Objectives**

**Meeting Contract Objectives:**
- In FY 2017, the RGCS program (with the inclusion of University of Iowa clinic sites) conducted 514 clinics in 8 Iowa communities, providing 2,749 clinic visits for patients and/or families (Figures 1, 2, & 3; Tables 3, 4 & 5). Of these, 45 clinics were held at regional outreach sites, 453 clinics were held at the University of Iowa Hospitals and Clinics, and 16 clinics were held via telemedicine with the provider located at the University of Iowa Hospitals and Clinics. Including all clinic locations, approximately 6,321 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 (31 of the 45) 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; 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 the Mechanisms of Health and Disease course 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, junior high, high school, and undergraduate students, the general public, and families. The genetics staff organizes an annual symposium for patients/families with Neurofibromatosis, a genetic
condition which predisposes individuals to the development of benign and/or malignant tumors along nerve tissue. They also host two education days (Davenport and Waterloo) for patients/families with Phenylketonuria (PKU), a metabolic condition requiring treatment with low protein foods and formulas. Additionally, a genetic counselor from the Division of Medical Genetics helped to organize the 2nd annual Iowa Statewide Sickle Cell Family Educational Symposium. Members of the Regional Genetic Consultation Service team have also done poster presentations at national genetics meetings including: the American College of Medical Genetics, the National Society of Genetic Counselors, the American Society of Human Genetics, and the Association of Public Health Laboratories Newborn Screening and Genetic Testing Symposium.

- RGCS staff participates in the activities/work groups of the Heartland Regional Genetics and Newborn Screening Collaborative.
- RGCS staff continues 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 fees 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 counselors in the 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 2017

#### National Performance Measure #11

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

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 a 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.

**National Performance Measure #12**

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.

The RGCS staff provides 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 assists 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 2,858 regional outreach clinics with more than 28,953 patient/family visits over the past 41 years, including 45 outreach clinics (427 patient visits) in FY17. In addition, 469 clinics (2,322 patient visits) were also held at UIHC and via telemedicine services in FY17. As the public’s knowledge and awareness of genetic services continue to expand, the referrals for genetic evaluations and services (for RGCS and UIHC clinics) continue to grow, exceeding the clinic and staff capacity. In May 2015, two genetic nurse/counselors retired, and clinic visits were decreased in the six 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. FY16 brought additional staffing challenges. While a new RGCS/UI genetics physician joined the staff in July 2015, two geneticists also left Iowa to take positions in other states in FY16. FY17 saw the departure of one and addition of another genetics physician. Two additional ARNPs/PAs were hired in FY17 to evaluate a specific patient population in the Connective Tissue Clinic, due to an average waitlist time of >18 months for new patients to be seen in this specialty genetics clinic at the time. Finding adequate numbers of qualified genetics personnel (physicians, LIPs, and counselors/nurses) continues to be a significant challenge. Recruitment for physicians and genetic counselors is ongoing.

The typical wait time for a genetics appointment for a “non-critical” patient is currently 12-15 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 inadequate. 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 (e.g. 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 have 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 sites, and 2nd party payer reimbursement are a few of the areas that have served as barriers to the establishment of 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. 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. 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, patients/families 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.

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. The Regional Genetic Consultation Service began 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 hereditable 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. Another example is the utilization of gene therapy for inherited forms of blindness. Researchers at the University of Iowa and elsewhere are developing treatments that are aimed at both preventing blindness and restoring vision for several genetic blinding disorders. Successful treatment of one specific childhood form of blindness has already been implemented and others are soon to be implemented.
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.