Regional Genetic Consultation Service

Contract 5882BD01
July 1, 2021 through June 30, 2022

Congenital & Inherited Disorders
Division of Health Promotion & Chronic Disease Prevention
Phone: 1-800-383-3826
http://idph.iowa.gov/genetics

Regional Genetic Consultation Service:
A contract established by IDPH in the Division of Health Promotion & Chronic Disease Prevention, Contract # 5882BD01
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
&
Stead Family Department of Pediatrics
Division of Medical Genetics & Genomics
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 & Genomics 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 birth defects and genetic disorders.

- Approximately 37,000 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 when considering the chances for 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 services are provided by a team of medical specialists, which includes Board Certified Medical Geneticists, Nurse Practitioners, Physician Assistants, Genetic Counselors, and Nurse Clinicians.

Common Types of Genetic & Congenital Disorders

Introduction to Genetic and Congenital Disorders:

A genetic disorder is a condition caused by an abnormality in an individual's DNA (deoxyribonucleic acid). Genes are the segments of DNA that provide instructions for the body on how to make certain proteins used in growth, development, function, and physical characteristics. Tightly wound DNA is 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, such as cleft lip and palate or spina bifida, are often multifactorial in nature, caused by a combination of environmental factors and one or more genetic changes. For more information: http://ghr.nlm.nih.gov/.

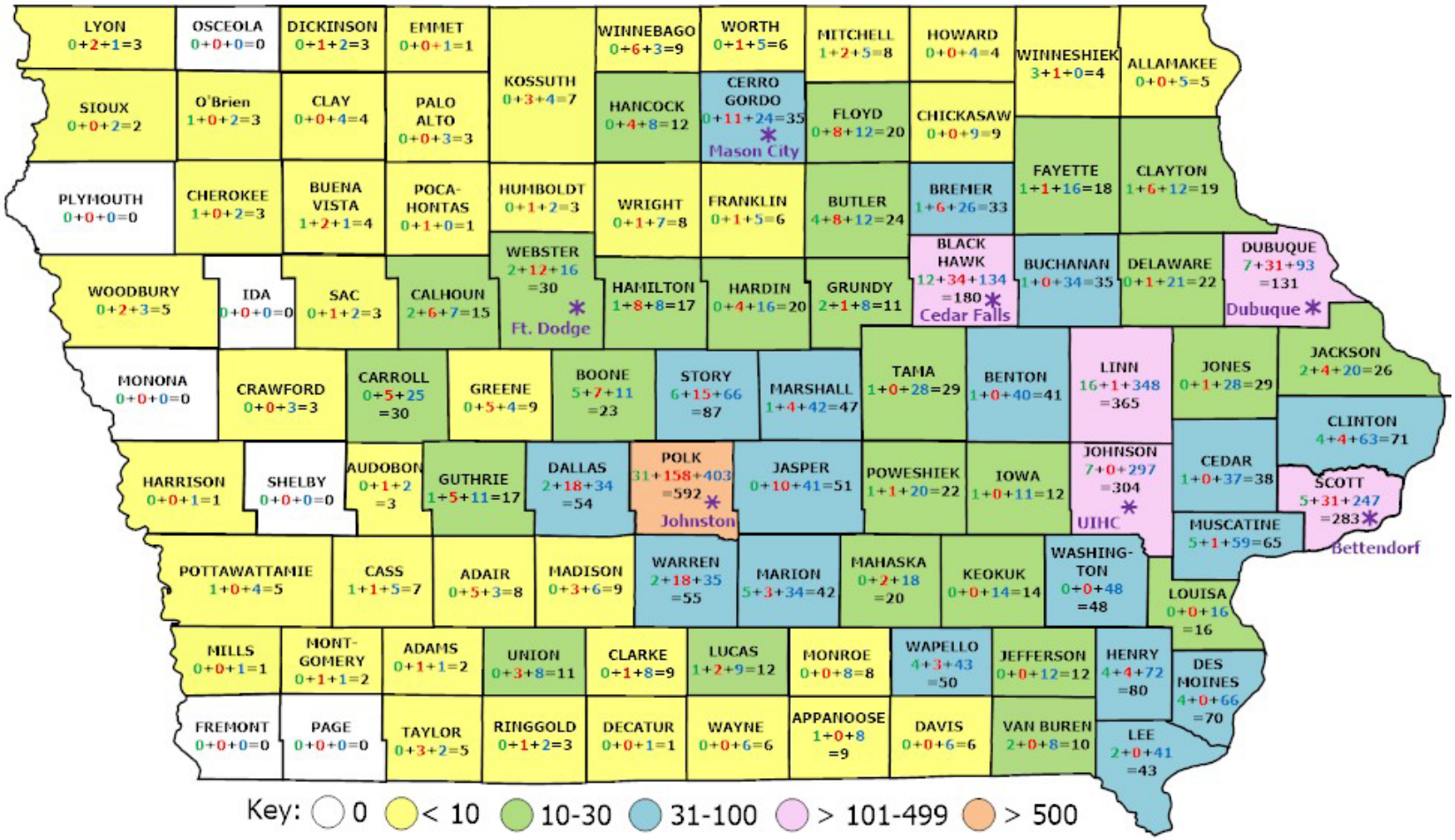
Clinical Subspecialty Services Provided

General Genetics	The general genetics clinic provides diagnostic evaluation, testing, counseling, and ongoing management for patients with known or suspected genetic conditions. Common reasons to be seen in this clinic include birth defects, learning or behavior concerns, problems with growth, family history of known genetic conditions, or concerns about a specific genetic condition.
Connective Tissue Disorders Clinic	The connective tissue disorders clinic provides diagnostic evaluation, testing, counseling, and ongoing management for children and adults with disorders affecting structural connective tissues. These include disorders of the skin, ligaments, tendons, and blood vessels. An individual may be referred to this clinic for diagnosis or ongoing management of the following conditions (as examples): Classic or vascular Ehlers Danlos Syndrome, Marfan syndrome, Loeys-Dietz syndrome, or Thoracic aortic aneurysm disorder.
Neurofibromatosis Clinic	The Neurofibromatosis (NF) clinic is a nationally affiliated member of the NF Clinic Network through the Children's Tumor Foundation. The clinic provides diagnosis and medical management to patients of all ages as well as access to a multidisciplinary care team. Primary conditions include neurofibromatosis types 1 and 2 (NF1 and NF2) and Schwannomatosis.
Metabolic Clinic	The metabolic genetics clinic provides diagnosis, education, medical treatment, management, and nutritional support for children and adults born with inherited disorders of metabolism. These conditions affect how the body makes or uses energy from foods. Common reasons for evaluation in this clinic include abnormal newborn screening, family history of a metabolic condition, poor growth or development, loss of skills, and unexplained changes to health status.
Cancer Predisposition Clinic	The cancer predisposition clinic provides diagnosis and management recommendations for adult and pediatric patients affected with or at-risk for genetic cancer predisposition syndromes. Common diagnoses include APC-associated polyposis syndrome, Beckwith-Wiedemann syndrome, PTEN-hamartoma tumor syndrome, ataxia telangiectasia, Birt-Hogg-Dube syndrome, von Hippel-Lindau syndrome (VHL), Juvenile Polyposis syndrome, SDH-related paraganglioma and pheochromocytoma syndromes to name a few. This clinic is nationally recognized by the VHL alliance.
Tuberous Sclerosis Complex Clinic	The multidisciplinary Tuberous Sclerosis Complex (TSC) clinic provides comprehensive care to patients with TSC. During a typical clinic visit, patients see a geneticist, genetic counselor, pediatric neurologist, pediatric nephrologist.
Lysosomal Storage Disease Clinic	The Lysosomal Disorders program evaluates children and adults with suspected or known lysosomal disorders such as Fabry Disease, Gaucher Disease, metachromatic leukodystrophy, mucopolysaccharidoses, and Niemann-Pick disease. These multisystem disorders may be diagnosed early in life (including via newborn screening) or much later in adulthood. In addition to offering regular clinical care (including management of enzyme replacement and other therapeutic options), the program participates in several clinical trials offering novel experimental therapies, including gene therapies, for these disorders.

Figure 1

FY 2022 Genetics Patient Census by County

(Telemed is shown in green, RGCS is shown in red; UIHC is shown in blue)



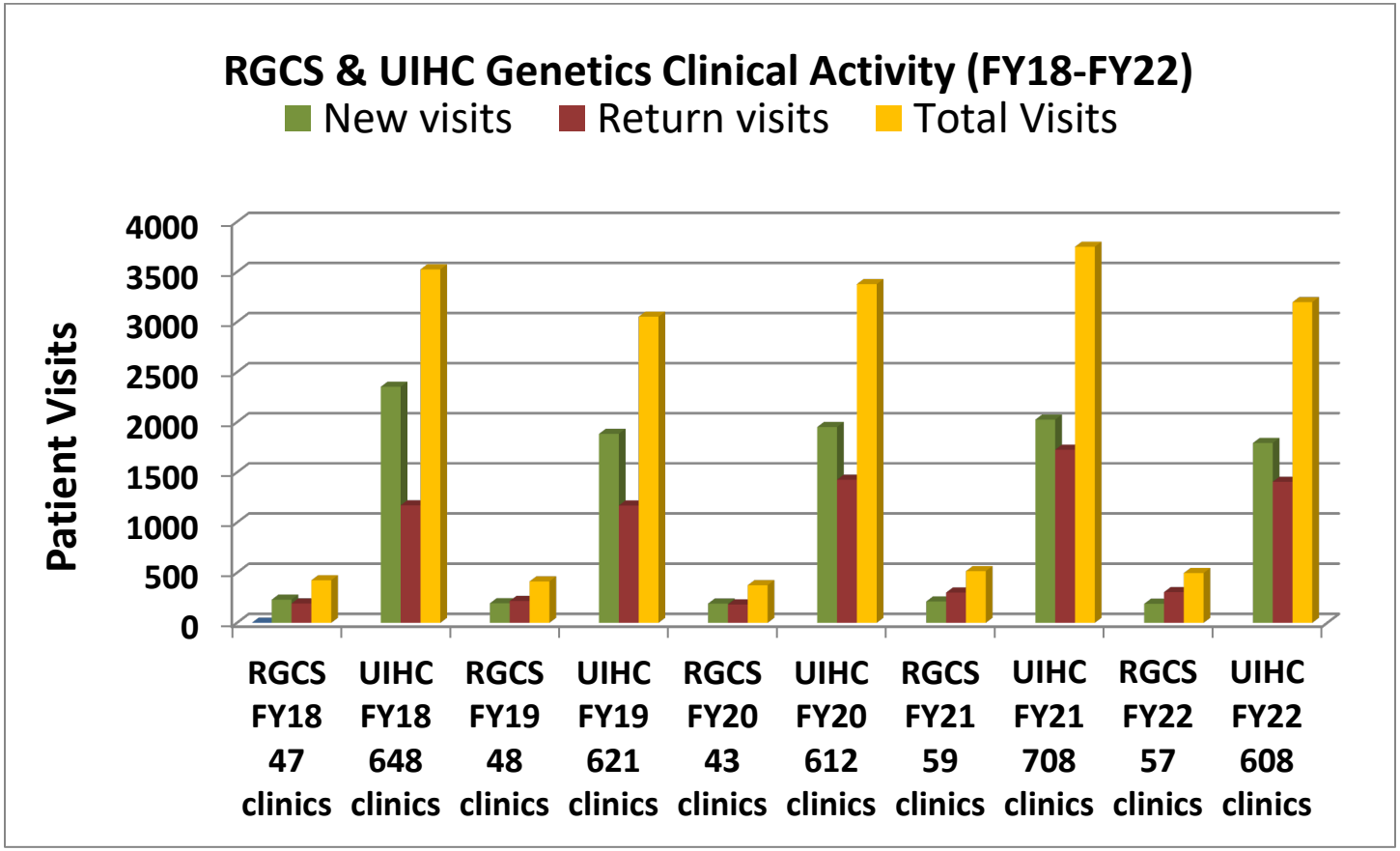
Total Patients = 3693; * Clinic Sites

Outliers: Total Patients = 201 (included in total)

201 patients from surrounding states were also seen by providers in the Division of Medical Genetics & Genomics in FY22

RGCS & UIHC Clinical Activity

Figure 2



The total number of UIHC clinics includes telemedicine clinics, held via a combination of both virtual telephone visits and virtual video visits.

In FY22, there were 187 full day clinics and 421 half day clinics held at UIHC or via telemedicine, in addition to 57 full day clinics held at six additional outreach locations across the state of Iowa.

In FY20, there were 53 scheduled outreach clinics. Due to the COVID-19 pandemic, 10 of these clinics were changed to telemedicine.

Table 1

RGCS & UIHC Genetics Clinical Activity FY18 – FY22					
Patient Visits	**RGCS & UIHC FY18 695 clinics**	**RGCS & UIHC FY19 669 clinics**	**RGCS & UIHC FY20 655 clinics**	**RGCS & UIHC FY21 767 clinics**	**RGCS & UIHC FY22 665 clinics**
New visits	2584	2078	1951	2025	1982
Return visits	1364	1387	1426	1725	1711
Total Visits	3948	3465	3377	3750	3693
Individuals present for counseling sessions	8915	7564	7478	7754	7677

**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. In FY20, visitor restrictions were put in place due to the COVID-19 pandemic. These restrictions limited the number of individuals present for clinic evaluations in both FY20, FY21, and FY22.

Combined RGCS & UIHC Total Patient Population by Age and Biological Sex

Table 2

Combined RGCS & UIHC Total Patient Population by Age and Biological Sex FY 22					
Age	**Female**	**Male**	**Female %**	**Male %**	**Total**
1-364 days	230	282	44.92%	55.08%	512
1-4 years	321	462	41.00%	59.00%	783
5-17 years	667	721	48.05%	51.95%	1388
18+ years	677	333	67.03%	32.97%	1010
Totals	**1895**	**1798**	**51.31%**	**48.69%**	**3693**

Services Provided by the Regional Genetic Consultation Service Per Contract

The RGCS contract for FY 2022 required a minimum of 135 regional genetic consultation clinics via on-site services or telehealth, providing at least 1,000 patient visits during the contract period. Expectations and services provided for individuals and families who have, are suspected to have, or are at risk for congenital and/or inherited disorders included:

Physician Evaluation and Medical Management

- **Consultation by board-certified medical 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/Case Management, 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 chances for recurrence, identification of at-risk relatives, and available reproductive options
  - 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/or 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 (i.e. Children with Special Health Care Needs community-based clinics, Early ACCESS)

Evaluation of RGCS FY 2022
RGCS Met Contract Objectives

Meeting Contract Objectives:

- In FY 2022, the RGCS program (with the inclusion of University of Iowa clinic sites) conducted:
  - 665 clinics (244 full day and 421 half day clinics) in 7 Iowa communities (Figures 1 & 2)
    - 57 clinics held at regional outreach sites
    - 594 clinics held at the University of Iowa Hospitals and Clinics
    - 14 clinics held via telemedicine with the provider located at the University of Iowa Hospitals and Clinics, although many other in person clinics included 1-3 telemedicine visits for patients unable to travel to on-site services. Altogether, 156 patient visits were completed via telemedicine in FY22.
  - 3,693 clinic visits for patients and/or families (Figures 1 & 2; Tables 1 & 2).
  - Including all clinic locations, approximately 7,677 patients, family members, and caregivers participated in these visits.
  - Of note, in addition to the 3,693 outpatient clinic visits, 299 genetic consults were completed during FY22 for patients from across the state of Iowa who were admitted to the Stead Family Children's Hospital or University of Iowa adult inpatient units.

Clinics were staffed by geneticists, nurse practitioners, physician assistants, and genetic 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 (Table 2). The majority of the regional outreach clinics (32 of the 57) were held in the Des Moines/Johnston site, with another 4 each 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, both at UIHC and external hospitals across the nation, residents, medical students, dietetic interns, high school students, 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.
- RGCS staff participates in the activities/workgroups of the Heartland Regional Genetics and Newborn Screening Collaborative.
- RGCS staff continues to participate in CIDAC meetings and activities. Two RGCS faculty and staff members currently hold leadership roles as voting members on CIDAC.
- 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.

The oversight of the organization and structure of the RGCS program is 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 & Genomics 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 & State Performance Measures / Priorities Outlined in Iowa's Family Health Plan that RGCS Participated in During FY 2022

National Performance Measure 6:
Percent of children, ages 9 through 35 months, who received a developmental screening using a parent-completed screening tool in the past year:

National Performance Measure 11:
Percent of child with and without special health care needs, ages 0-17, who have a medical home:

National Performance Measure 12:
Percent of adolescents with and without special health care needs, ages 12-17, who received services to prepare for the transition to adult health care:

National Outcome Measure 17.1, 17.3, 17.4:
To track the percent of children and youth with special health care needs, autism spectrum disorder (ASD), and attention deficient disorder/attention deficit hyperactivity disorder (ADD/ADHD)

National Outcome Measure 17.2:
To ensure access to needed and continuous systems of care for children and youth with special health care needs

National Outcome Measure 21:
Percent of children ages 0-17 without health insurance; To ensure access to needed health care services for children

Children are considered to have a special health care need if, in addition to a chronic medical, behavioral, or developmental condition that has lasted or is expected to last 12 months or longer, they experience either service-related or functional consequences, including the need for or use of prescription medications and/or specialized therapies. The percent of children with special health care needs has been increasing since 2001. About 1 in 5 of all US children are considered to have special health care needs. However, they account for almost half of all health care expenditures for children (U.S. Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau 2014). During genetics evaluations, developmental screening questions are utilized to determine if a child may benefit from a referral for a more comprehensive developmental evaluation or referrals to services such as Early ACCESS.

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. 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. While individuals across the lifespan are cared for in the RGCS and UIHC Genetics clinics, genetics staff do discuss and assist with referrals as needed to transitional programs or adult health care subspecialists for those individuals with special health care needs who are preparing for adulthood.

As part of its role with families and patients, the genetics staff assists families/parents to identify social services and financial resources to meet their health care needs, aiding in the application process through referrals to social work, if needed. Staff advocates for patients by communicating with health insurers regarding the importance of appropriate diagnostic testing, management and treatment for specific (often rare) genetic disorders, providing educational resources as needed. Genetics 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 Medicaid Clinical Advisory Committee regarding health care needs of children/patients with complex health conditions.

References:
https://mchb.tvisdata.hrsa.gov/PrioritiesAndMeasures/ListOfStatePerformanceMeasures
https://mchb.tvisdata.hrsa.gov/PrioritiesAndMeasures/NationalOutcomeMeasures
https://mchb.tvisdata.hrsa.gov/PrioritiesAndMeasures/EvidenceBasedStrategyMeasures/1

Barriers, Challenges and Additional Facts

The RGCS program has held 3,112 regional outreach clinics with more than 31,500 patient/family visits over the past 46 years, including 57 full day outreach clinics (496 patient visits) in FY22. In addition, 187 full day and 421 half day clinics (3,197 patient visits) were also held at UIHC and via telemedicine services in FY22

Challenge #1: Patient access due to nationwide shortage of clinical genetics providers
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. This is not a challenge unique to Iowa. According to the American Board of Medical Genetics and Genomics and the National Society of Genetic Counselors, in 2019, there were approximately 1,760 board certified clinical geneticists and approximately 5,170 board certified genetic counselors. Nationwide, this equates to a ratio of approximately 1 clinical geneticist to every 186,000 individuals in the general population and 1 genetic counselor to every 63,000 individuals in the general population. In Iowa, the clinical provider: population ratio for FY22 was 1 clinical geneticist to every 455,000 individuals and 1 clinical genetic counselor to every 131,000 individuals.

Over the past several years, scheduled clinics have been altered in accordance with new or departing faculty and staff. As examples:

- FY17 –Departure of one genetic counselor and one geneticist/Retirement of one genetics-nurse counselor/Addition of three genetic counselors and one metabolic nurse/Addition of one ARNP and one PA to evaluate a specific patient population in the Connective Tissue Disorder Clinic, due to an average waitlist time of >18 months for new patients to be seen in this specialty genetics clinic at that time.
- FY18 – Departure of one geneticist; Addition of one geneticist and one metabolic nurse
- FY19 – Departure of one ARNP, one metabolic nurse; Addition of one geneticist
- FY20 – No departures or additions
- FY21 – Addition of one geneticist and one genetic counselor
- FY22 – Departure and addition of one genetic counselor; Departure of one nurse practitioner. Addition of genetic counseling only clinics.

Finding adequate numbers of qualified genetics personnel (geneticists, LIPs, and genetic counselors/nurses) continues to be a significant challenge. Recruitment for medical geneticists is ongoing.

The typical wait time for a genetics appointment for a “non-critical” patient is currently 6-9 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:

1) Timely review and assessment of every incoming patient referral for acuity
2) Prioritization of patient/physician requests for clinic appointments
3) Increasing the number of patient appointments per clinic day
4) Overbooking appointments at the University on non-clinic days when needed and
5) Providing medical recommendations for local providers to begin the evaluation process prior to the genetics appointment.

Furthermore, significant efforts were made in FY18-FY20 and again in FY22 to increase the number of “genetic counseling only” and ARNP/PA clinics. This action allowed those patients who have established diagnoses with published medical management guidelines or patients not requiring a physical examination or ongoing medical management by a medical geneticist to be seen in a much timelier fashion. This action in part allowed the general genetics waitlist to be decreased from 12-15 months to 6-9 months for the first available “non-critical” opening.

Challenge #2: Transportation

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) are utilized when possible. 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. In FY19, clinical schedules were altered to provide added clinic appointments to the Fort Dodge and Mason City outreach clinics. 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.

After much effort over the years, the COVID-19 pandemic did result in expanded use of telemedicine technology and services. At the end of FY22, the first recurring telemedicine clinic staffed by a medical geneticist and genetic counselor was implemented. This included expansion of telemedicine services to four UIHC hub sites in Cedar Falls, Bettendorf, Johnston, and Dubuque. We look forward to continuing and expanding these telemedicine clinics in order to remove barriers to access in the years to come. While telemedicine appointments allow for increased access for patients with certain clinical indications, there are populations of patients that require dedicated physical exams which must be completed through an in person visit. In addition, barriers for reimbursement of genetic counseling services when using telemedicine continues to be an ongoing issue.

Challenge #3: Insurance barriers

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 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. In other instances, insurance policies place constraints on testing modality/type of testing that can be considered for a patient's care. 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.

Challenge #4: Funding

With continued statewide budget constraints, funding for this program has either decreased or remained neutral in each of the previous five years. Along with rising salaries needed to retain skilled clinicians in high demand, budget cuts have strained this program to provide the same high level of care with fewer resources. A consistent funding source with appropriate yearly increases is needed for the long-term sustainability and success of the RGCS program.

Regional Genetic Consultation Service Director Statement

Alpa Sidhu, MBBS, PhD
Clinical Associate Professor of Pediatrics

Medical genetics and genomics 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 and genomics focuses not solely on individual patients, but on the entire family.

Obtaining 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 an underlying genetic diagnosis, education about the condition with resources, surveillance, recurrence risk and management based on current practice guidelines.

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.

Lysosomal storage disorders are a group of conditions that result in accumulation of storage material leading to disease. Treatment is now available for a number of these conditions that would change morbidity and mortality for these patients.

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.