Regional Genetic Consultation Service

Contract 5880BD01
July 1, 2019 through June 30, 2020

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 # 5880BD01
Administrative Code, Chapter 4:641-4.5(136A)

Iowa Department of Public Health
Advancing Health Through the Generations

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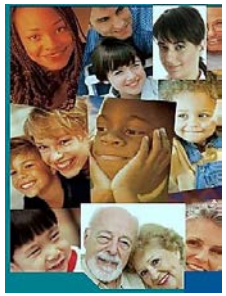

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In collaboration with the
Iowa Department of Public Health
&
The Stead Family Department of Pediatrics
Division of Medical Genetics & Genomics
University of Iowa Stead Family Children's Hospital
&
Clinical Outreach Services
University of Iowa Hospitals 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 congenital defects and genetic disorders.

- Approximately 37,500 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 services are provided by a team of medical specialists, which includes Board Certified Medical Geneticists, Nurse Practitioners, Physician Assistants, Genetic Counselors, and Nurse Clinicians.

A Patient Story

Ethan really enjoys going to school, where he has taken great interest in books. His love for music started when he was just an infant and has continued to help him in so many ways today. You can be sure to find on his playlist some Sir Paul McCartney! He has been a fan ever since his dad put the needle on the record of the album, "McCartney."

*Verbal and written permission were obtained from Ethan's parents to include his photo and story in this report. Family supplied the photo. We thank Ethan's family for the opportunity to highlight their incredible son.

After much difficulty with feeding and weight gain early in life along with some delays in development, Ethan's geneticist at UIHC noted his broad thumbs on physical exam. Completed genetic testing confirmed his suspected diagnosis of Rubinstein-Taybi syndrome (RSTS). This diagnosis allowed his care team to outline specific recommendations for his ongoing medical care.

Introduction: ***Rubinstein-Taybi syndrome (RSTS)***

- A genetic disorder characterized by short stature and difficulty with gaining weight, intellectual disabilities, and broad thumbs and broad first toes
- Cause: a change or mutation in any one of two different genes can cause RSTS. Approximately 30% 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 RSTS.
- Affects approximately 1:100,000 individuals. Mild forms of the disorder are likely to be overlooked and underdiagnosed.
- Reported in all races and socioeconomic backgrounds.
- Most individuals with RSTS have no family history of the condition. Rather, the condition occurs as a new spontaneous change in the individual for the first time.

Features and Characteristics:

Associated symptoms and findings may vary greatly in range and severity. However, common features include:

- Short stature
- Difficulty gaining weight
- Developmental delay and intellectual disabilities of variable degrees
- Broad (wide) thumbs and broad first toes
- Less commonly, extra fingers or toes, bifid or angled thumbs or toes
- Structural heart defects

- Structural kidney defects in males and females and/or undescended testes in males
- Characteristic facial features such as down-slanting eyes, nasal tip extending below the nares, low set ears, high arched palate, talon cusps (tooth finding), extra or missing teeth
- Hormone disorders (growth hormone deficiency, low thyroid levels)
- Gastrointestinal problems (acid reflux, constipation, Hirschsprung disease)
- Eye findings (cataracts, coloboma, glaucoma, “lazy eye”)
- Possible increased risk for cancer (however, no specific surveillance recommendations exist currently)

Diagnosis:
The diagnosis of RSTS can often be made by a medical geneticist through physical exam and review of clinical history. Genetic testing can be helpful in confirming the diagnosis. Medical management guidelines for individuals with RSTS were published in 2015, which can be used to guide care and surveillance.

Treatment:
There is no cure for RSTS, 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 RSTS. 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:

https://www.eurordis.org/living-with-a-rare-disease

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). DNA is the molecule that contains the genetic code of living things. 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)
- 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/.

Table 1

Examples of Genetic/Congenital Disorders & Reasons for Referral	
***This list is meant to provide examples and is not all-inclusive**	
Angelman syndrome	Lesch-Nyhan syndrome
Apert syndrome	Lissencephaly
Beckwith-Wiedemann syndrome	Marfan syndrome
CHARGE syndrome	Mitochondrial Disorders
Cleft lip and / or Palate	Neurofibromatosis
Cornelia de Lange syndrome	Noonan syndrome
Developmental / Growth Delay	Phenylketonuria (PKU)
Down syndrome	Prader-Willi syndrome
Dysmorphic facial characteristics	Rett Syndrome
Ehlers Danlos syndromes	Rubinstein-Taybi syndrome
Fabry Disease	Sanfilippo syndrome
Familial Cancer Syndromes	Septo-Optic Dysplasia
Fetal Alcohol Syndrome	Smith-Lemli-Opitz syndrome
Fragile X syndrome	Smith-Magenis syndrome
Galactosemia	Sotos syndrome
Hereditary Hemorrhagic Telangiectasia	Spina bifida
Hunter syndrome	Trisomy 13 or 18
Hydrocephalus	Tuberous Sclerosis
Kabuki syndrome	Turner syndrome
Klinefelter syndrome	Williams syndrome
Leigh 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		
Genetic/Congenital Disorders	***Incidence Of Genetic / Congenital Disorders***	***Estimated # Newborns affected***
Congenital Malformations (Newborns)	~4%	1,500/yr
• Chromosome Abnormalities	~1%	400/yr
• Multifactorial/ Single gene / Environmental	~3%	1,100/yr
Other Diseases/disorders with Genetic components	***Incidence or Prevalence***	***Estimated # Affected***
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.

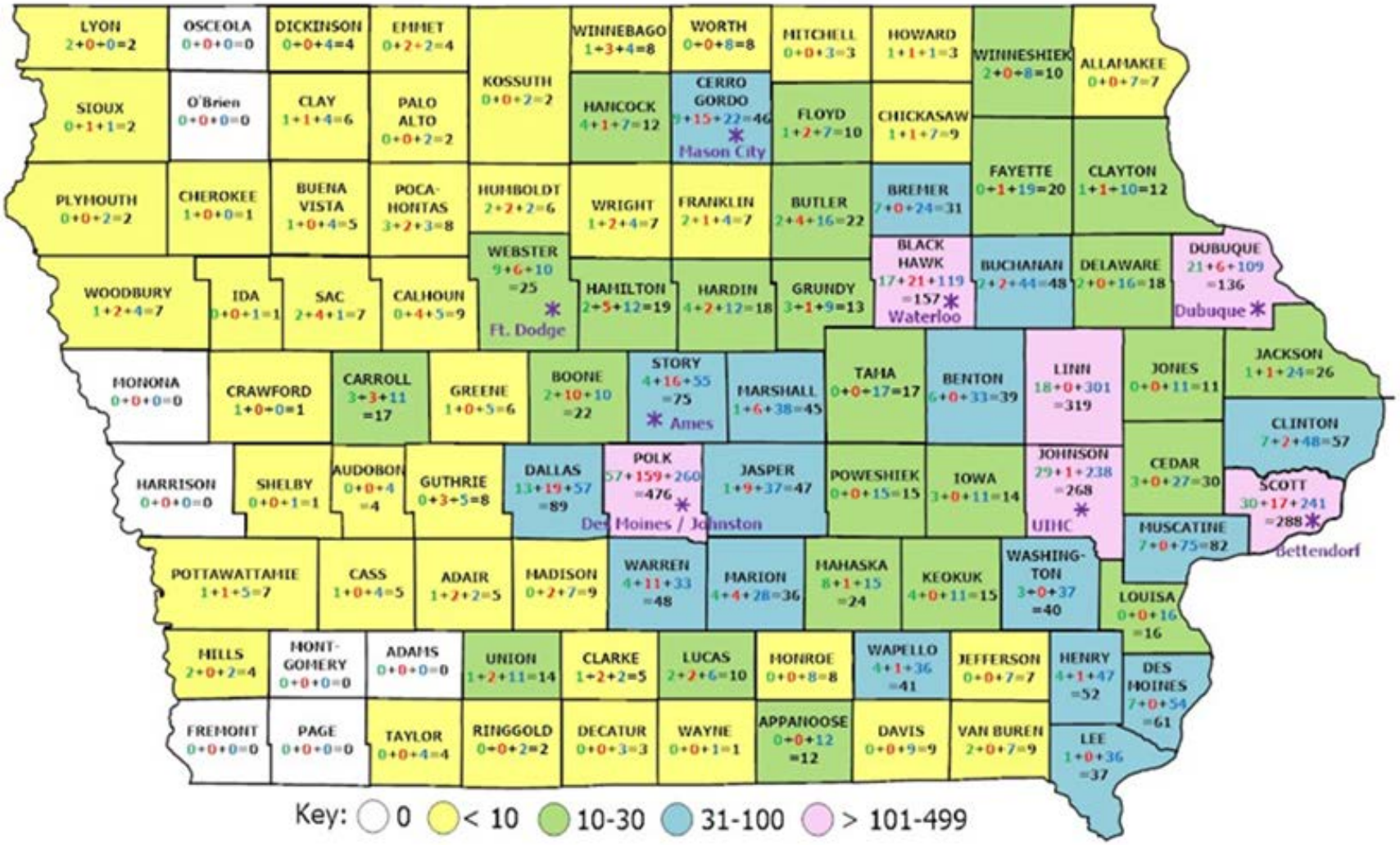
References:

https://tracking.idph.iowa.gov/Health/Birth-Defects/About-Birth-Defects-Data
http://www.cancer-rates.info/ia
https://idph.iowa.gov/Portals/1/userfiles/35/2019_IRCID%20Report%20Final.pdf

Figure 1

FY 2020 Telemedicine, RGCS, & UIHC Genetics Patient Census by County

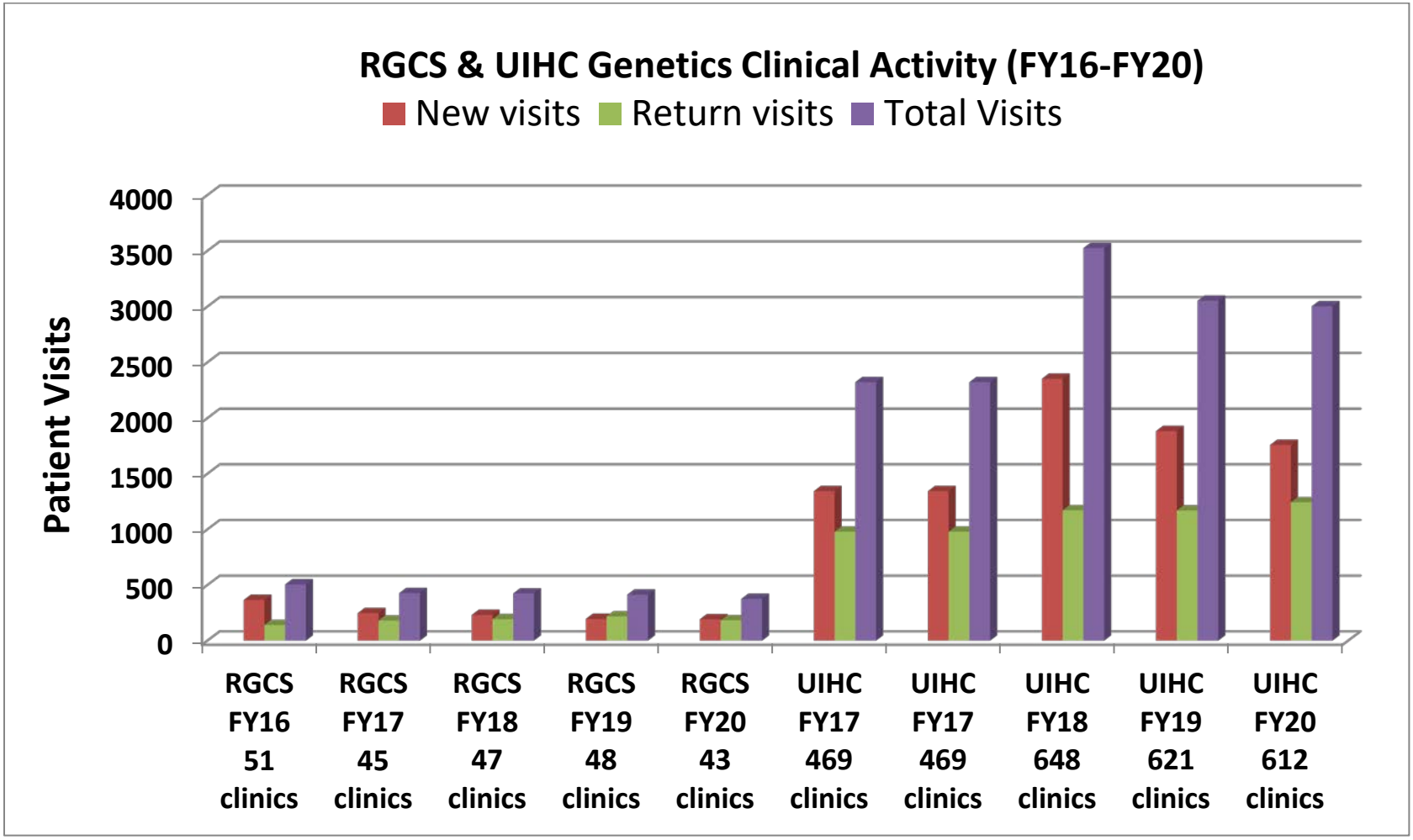
(Telemedicine is shown in green; RGCS is shown in red; UIHC is shown in blue)



Outliers: Total Patients = 187 (included in total)
187 patients from surrounding states were also seen by providers
in the Division of Medical Genetics & Genomics in FY20

RGCS & UIHC Clinical Activity

Figure 2



In FY20, there were 53 scheduled RGCS clinics. Due to the COVID-19 pandemic, 10 of these clinics were changed to telemedicine. The total number of UIHC clinics includes telemedicine clinics, held via a combination of both virtual telephone visits and virtual video visits. Multiple UIHC clinics were also cancelled while appropriate personal protective equipment was obtained, thus reflecting an overall decrease in number of clinics held in FY20 as compared to previous years.

Table 3

RGCS & UIHC Genetics Clinical Activity
FY16 – FY20

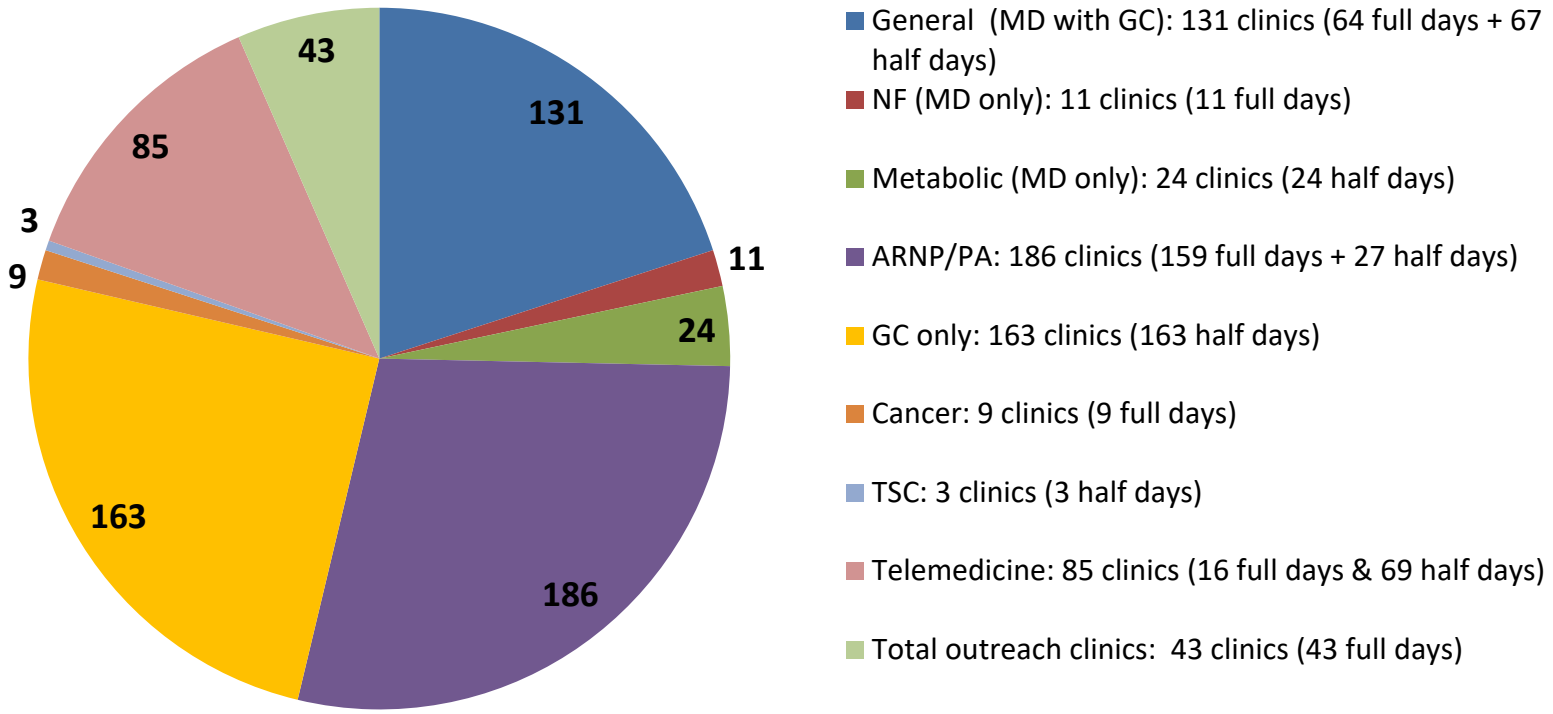
Patient Visits	RGCS & UIHC FY16 467 clinics	RGCS & UIHC FY17 514 clinics	RGCS & UIHC FY18 695 clinics	RGCS & UIHC FY19 669 clinics	RGCS & UIHC FY20 655 clinics
New visits	1096	1590	2584	2078	1951
Return visits	1279	1159	1364	1387	1426
Total Visits	2375	2749	3948	3465	3377
Individuals present for counseling sessions	NA	6321	8915	7564	7478

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

**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 counseling sessions.

Total RGCS & UIHC Clinics (MD, PA, ARNP, GC only) FY20

Figure 3



*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 (FY16 – FY20)

Table 4

	FY 2016		FY 2017		FY 2018		FY 2019		FY 2020		% Gender by Age (5 year average)		
AGE	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	
1-364 dys	22	34	30	25	24	24	17	23	17	15	5.14%	5.74%	
1-4 yrs	49	72	74	51	63	73	63	82	54	67	14.15%	15.50%	
5 YEAR TOTAL UNDER 5 YEARS of AGE													**41%**
5-9 yrs	53	66	48	39	33	56	39	51	37	47	9.80%	11.62%	
10-14 yrs	49	45	29	42	35	35	36	31	32	23	8.45%	8.64%	
15-19 yrs	32	16	21	19	21	21	11	18	20	26	4.90%	4.39%	
5 YEAR TOTAL 5-19 YEARS of AGE													**48%**
20-29 yrs	19	9	12	13	7	10	9	5	7	6	2.52%	2.05%	
30-39 yrs	17	4	6	2	8	3	13	3	6	4	2.33%	0.84%	
40-49 yrs	6	3	6	4	2	3	3	0	4	1	0.98%	0.65%	
50-59 yrs	7	2	5	0	2	1	2	0	4	1	0.93%	0.33%	
60-69 yrs	0	0	1	0	1	0	4	3	2	2	0.37%	0.23%	
70 &older	0	0	0	0	0	0	0	0	0	0	0.00%	0.00%	
5 YEAR TOTAL OVER 20 YEARS of AGE													**11%**
Totals	**254**	**251**	**232**	**195**	**196**	**226**	**197**	**216**	**183**	**192**	**1062**	**1080**	**100%**

Combined RGCS & UIHC Total Patient Population by Age and Gender (FY16 – FY20)

Table 5

	FY 2016		FY 2017		FY 2018		FY 2019		FY 2020		% Gender by Age (5 year average)		
AGE	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	**Female**	**Male**	
1-364 dys	114	190	111	145	150	179	164	220	176	216	4.63%	5.89%	
1-4 yrs	196	252	266	274	332	358	339	423	269	331	9.08%	10.21%	
5 YEAR TOTAL UNDER 5 YEARS of AGE													**30%**
5-9 yrs	203	236	239	233	277	342	250	305	238	289	7.82%	8.77%	
10-14 yrs	189	189	205	220	345	275	259	257	204	200	7.78%	7.42%	
15-19 yrs	146	112	205	129	324	225	199	184	193	145	6.91%	5.46%	
5 YEAR TOTAL 5-19 YEARS of AGE													**44%**
20-29 yrs	131	69	183	79	248	67	208	87	154	90	5.98%	2.95%	
30-39 yrs	113	38	151	36	279	52	150	71	158	75	5.51%	2.30%	
40-49 yrs	72	28	105	37	148	35	113	51	79	36	3.35%	1.49%	
50-59 yrs	53	15	75	18	91	34	67	27	53	26	2.20%	0.95%	
60-69 yrs	24	7	26	8	56	18	44	25	33	20	1.19%	0.59%	
70 &older	1	0	3	1	8	3	11	11	12	5	0.23%	0.17%	
5 YEAR TOTAL OVER 20 YEARS of AGE													**27%**
Totals	**1242**	**1136**	**1569**	**1180**	**2258**	**1588**	**1804**	**1661**	**1569**	**1433**	**8442**	**6998**	**101%**

Services Provided by the Regional Genetic Consultation Service Per Contract

The RGCS contract for FY 2020 required a minimum of 135 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 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 (i.e. Children with Special Health Care Needs community-based clinics, Early ACCESS)

Evaluation of RGCS FY 2020
RGCS Met Contract Objectives

Meeting Contract Objectives:

- In FY 2020, the RGCS program (with the inclusion of University of Iowa clinic sites) conducted:
  - 655 clinics in 7 Iowa communities (Figures 1, 2, & 3)
    - 43 clinics held at regional outreach sites
    - 527 clinics held at the University of Iowa Hospitals and Clinics
    - 85 clinics held via telemedicine with the provider located at the University of Iowa Hospitals and Clinics.
  - 3,377 clinic visits for patients and/or families (Figures 1 & 2; Tables 3, 4 & 5).
  - Including all clinic locations, approximately 7,478 patients, family members, and caregivers participated in these visits.

  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.

  The COVID-19 pandemic resulted in the closure of multiple UIHC on-site clinics in order to obtain proper personal protective equipment, the conversion of 10 RGCS clinics to telemedicine, the overall expansion of telemedicine, and a restriction on the number of visitors allowed to partake in clinic appointments.

- 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 43) were held in the Des Moines/Johnston site, 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; high school students; the general public; and families. RGCS staff planned educational activities for various STEM (Science, Technology, Engineering, and Math) events throughout the year. 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. Additionally, genetic counseling staff in the Division of Medical Genetics & Genomics participated in the 4th annual Iowa Statewide Sickle Cell Family Educational Symposium.
- 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.
- 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 2020

National Performance Measure / State Performance Measure #2

Percentage of children 0-21 served by Title V who meet Iowa's title V criteria as having a medical home.

National Performance Measure / State Performance Measure #6

Percent of CYSHCN with parents who are very satisfied with the communication among doctors and other health care providers.

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.

References:
https://mchb.tvisdata.hrsa.gov/PrioritiesAndMeasures/ListOfStatePerformanceMeasures
https://www.idph.iowa.gov/Portals/1/userfiles/142/State%20Performance%20Measures%20FFY2020.pdf

Barriers, Challenges and Additional Facts

The RGCS program has held 2,996 regional outreach clinics with more than 30,165 patient/family visits over the past 44 years, including 43 outreach clinics (375 patient visits) in FY20. In addition, 612 clinics (3,002 patient visits) were also held at UIHC and via telemedicine services in FY20.

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 2017, there were approximately 1,600 board certified clinical geneticists and approximately 4,600 board certified genetic counselors. Nationwide, this equates to a ratio of approximately 1 clinical geneticist to every 204,000 individuals in the general population and 1 genetic counselor to every 71,000

individuals in the general population. In Iowa, the clinical provider: population ratio for FY20 was 1 clinical geneticist to every 631,000 individuals and 1 genetic counselor to every 143,000 individuals.

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

- FY16 –Departure of two geneticists/Addition of two geneticists and one genetic counselor
- 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

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 3-6 months. Please note, however, that this is an improvement from the previous 6-9 month wait list a year ago. 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 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 3-6 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. The number of telemedicine clinics increased from 22 clinics in FY19 to 85 clinics in FY20. The first UI-Johnston telemedicine clinic staffed by a medical geneticist and genetic counselor was held in the Fall of 2019. Plans are being laid to expand telemedicine services to the additional UIHC hub-sites in Cedar Falls, Bettendorf, and Dubuque. We anticipate the creation of a recurrent medical geneticist/genetic counselor telemedicine clinic to begin mid FY21. 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 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 may be 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.

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

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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.