



MSRGN STATE TEAMS MEETING
UPP PROJECT
WEDNESDAY JAN 10, 2018



Our Identified Program Priorities

- Link medically underserved populations to genetic services;
- Implement quality improvement activities to increase genetic services to the medically underserved;
- Implement telehealth/telemedicine focused on clinical genetics;
- Distribute resources to providers (genetics & primary care), public health professionals, and individuals with genetic conditions and their families.

The challenge is developing a **service infrastructure** throughout our region **to facilitate access** to services for the underserved, creating **enduring solutions** in our states.

Underserved Patient Populations Project (UPP)

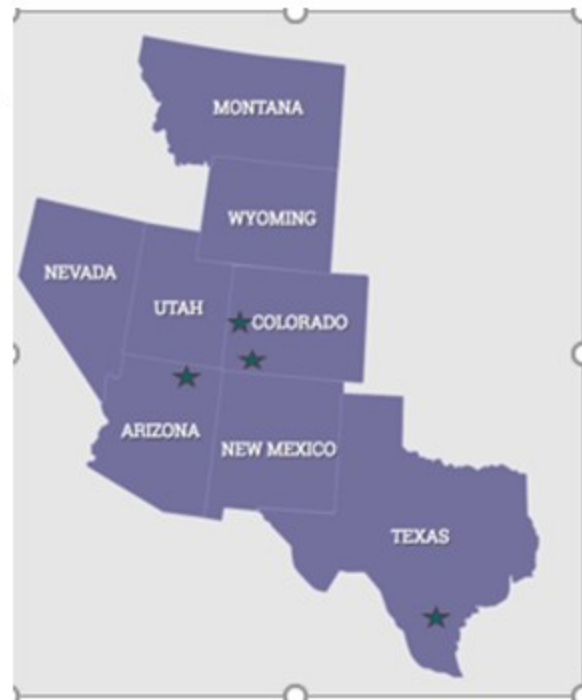
- Identify barriers in accessing genetic services at four initial sites
- Collect data on community characteristics available through Census and HRSA sources
- Review literature on access to care in American Indian and Hispanic populations
- Interview providers and families to clarify community barriers to genetic services
- Design interventions based on literature review and surveys
- Vet interventions with clinic directors
- Implement interventions in 4 communities
- Evaluate impact of interventions on patient access

Underserved Populations: Initial Target Sites for Y1

Through previous work and input from state leaders, MSRGN identified sites at which our Y1 efforts are focused:

- Tuba City, AZ
- Grand Junction, CO
- Durango, CO
- San Antonio, TX

Sites were selected because of participation of genetics providers, poverty levels, rural geographic location, and/or populations that experience health disparities



Identification of Barriers to Access

Review publicly available data sources (Anderson Model)

Review relevant literature

Survey providers and consumers

Select barriers that are amenable to intervention at the clinic and/or community level



Anderson Behavioral Model

Used to identify modifiable and non-modifiable factors that impact access to care

Selects community-level variables representing social, economic, structural, and public policy environments that influence use of medical care

Included in our analysis: community demographics, FQHCs, health care market factors

Davidson PL, Anderson RM, Wyn R, Brown ER. A Framework for Evaluating Safety-net and Other Community-Level Factors on Access for Low-Income Populations. *Inquiry* 41:21-38, 2004.

Community Data Collected

County-level demographic and socioeconomic data extracted from US Census Bureau sources

- Per cent low income
- Per cent Medicaid
- Per cent immigrant

Health system level resources identified from HRSA data sources

- FQHCs
- Population served by FQHCs



Collect demographic data related to barriers to care

Census Topic	Coconino
Total Population	
Population Estimates (7/1/16)[V2016]	140,908
Age	
Persons under 5 years, percent (7/1/16)[V2016]	5.80%
Persons under 18 years, percent (7/1/16)[V2016]	21.20%
Persons 65 years and over, percent (7/1/16)[V2016]	11.90%
Race and Hispanic Origin	
White alone, not Hispanic or latino percent (7/1/2016)[V2016]	54.70%
Hispanic or Latino, Percent (7/1/2016)[V2016]	13.80%
Black or African American alone, percent (7/1/2016)[V2016]	1.40%
American Indian and Alaskan Native alone, percent (7/1/2016)[V2016]	27.50%
Asian alone, percent (7/1/2016)[V2016]	1.90%
Native Hawaiian and other Pacific Islander alone, percent (7/1/2016)[V2016]	0.20%
Two or more races, percent (7/1/2016)[V2016]	2.80%
Population Characteristics	
Foreign Born Persons, percent (2011-2015)	5.40%
Families and Living Arrangements	
Language other than English spoken at home, percent of persons age 5+ years (2011-2015)	24.30%
Health	
With a disability, under age 65 years, percent (2011-2015)	8.20%
Persons without health insurance, under age 65 years, percent	14.60%
Economy	
In civilian labor force, total, percent of population age 16 years+ (2011-2015)	64.60%
Income and Poverty	
Median household income (in 2015 dollars) (2011-2015)	\$50,234
Per capita income in past 12 months (in 2015 dollars) (2011-2015)	\$24,308
Persons in poverty, percent	19.50%
Businesses	
Total employment, 2015	50,051
Geography	
Population per square mile, 2010	7.2
HRSA FQHC	
Number of FQHC hospitals in county	3
Location of FQHCs and total patients served (as of 2015)	CANYONLANDS COMMUNITY HEALTH CARE 827 Vista Ave, Page, AZ 86040 Total Patients Served: 17,368
	NORTH COUNTRY HEALTHCARE, INC. 2920 N Fourth St, Flagstaff, AZ 86004 Total Patients Served: 48,718
US Census Quick Facts Data	TUBA CITY REGIONAL HEALTH CARE CORPORATION 167 Main St, Tuba City, AZ 86045 Total Patients Served: 2,176
HRSA Health Center Program Websites	

Provider Surveys (n=8)

What types of genetics patients does your clinic see?

Does your practice/organization maintain a genetics patient waiting list? (Yes/No)

[if yes] Can you estimate how many patients are currently on that waiting list?

Excluding referrals of newborns with positive screens, what are patients and referring providers told is the current wait time for an initial genetics appointment? (probe for specifics
– months and weeks)



Do you feel like your clinic is able to meet the current clinical genetics need in your community/area? (Yes/No)

You said _____ made it difficult to meet current genetics service needs in your community. What could help your practice overcome this barrier?

Do you think there is a shortage of genetics providers to meet current clinical need in your area? (note that they may have mentioned this is the previous question)

What types of providers are in short supply? (probe for Pediatric clinical geneticist, Adult clinical geneticist, Metabolic geneticist, Molecular geneticist, Genetic counselor)

Do you think there are underserved individuals in your area who would benefit from genetic services but have difficulty getting them?

What are the major barriers for these individuals or families to receive genetic services? (probe for Geography (too far from clinics), Poor or no insurance coverage, Poor provider education on need for genetic services, Poor public education on benefits of genetic services, Low priority in relation to other medical needs of the patient, Long wait time for an appointment)

You said _____ is a barrier for individuals and families seeking or needing genetic services. What do you think would help families overcome this barrier?

Do Primary Care Providers assist in the evaluation and follow-up care of your patients?

How? Could they do more? What do they need in order to do more?

Who are the key partners you rely on for giving families resources, information, and support? (probe for national educational groups, support groups, community organizations, etc – get specific names)

What, specifically, does this partner provide?

Are there organizational and/or administrative barriers in your clinic that make it less efficient than it could be?

What are they? What would be needed to address these barriers?

Do you offer telegenetics services? Yes/No

Are those telegenetics services sufficient to meet patient needs? Do you feel there is room for expansion? What would your clinic need to fully expand telegenetics services? Do you have adequate institutional support to fully implement your telegenetics program? Is institutional privileging a barrier? Do you deliver any services across state lines and, if so, is licensing a barrier?

Would it help to add them? What would be necessary to add them?

What are the top two changes you would recommend your clinic undertake to facilitate provision of better access to genetic services in your community?

What resources would you need to make this change? (probe for financial and human resources, as well as training and information)

If you had all the resources you needed, how long do you think it would take to make the change?

How would you measure success of this change?

Consumer Surveys

6 consumers reviewed family survey questions and answered many of them

2 additional consumers from the target communities were interviewed using the finalized survey form



Please share your experiences with trying to find information or services for yourself or your children with learning problems, developmental delay, birth defects, or other special medical needs (for example, Down Syndrome). [CUSTOMIZE IF POSSIBLE]

How did your journey begin?

Does your child have a specific diagnosis? If yes, what is their diagnosis?

Can you describe your most recent visit to a genetics clinic? What went well in that appointment? What did not? Was this a typical visit? If not, can you describe a typical visit?

Thinking back to your initial visit to genetics, how long did you wait for an appointment? Did you have to travel to get to that appointment? What was it like being told your child might have some sort of genetic disorder and then having to wait for an appointment?

What is the biggest challenge your family has faced in accessing genetic services?

Have you ever experienced financial barriers to obtaining genetic services? (for example, your co-pay is too high or your insurance does not cover something and you are unable to pay out-of-pocket) What about genetic testing?

What information is/was helpful to know from your child's doctor? Is this information enough for you? Is it too much information?

Please explain.

Where do/did you look for and find information related to your or your child's genetic condition (for example, Down Syndrome)?

How do you find out about where to look for this information?

Did you find any useful information on the internet? What about social media?

What barriers or challenges exist when attempting to find information related to genetic conditions?

Are there support groups, other families, or resources available in your community? Do you interact with any of them? What types of information or resources do they provide to you?

What types of support would be helpful for you and your family?

Would you be interested in connecting with support groups? If yes, offer Family-to-Family Information Center, others.

When you first realized that you or your child had a special medical need [CUSTOMIZE IF POSSIBLE], to whom did you look , and where did you go, for the services your family needed?

What were the major barriers or challenges you faced when *initially* accessing genetic services (e.g. seeing a geneticist, getting genetic testing)? (probe for Geography (too far from clinics), Poor or no insurance coverage, Poor provider education on need for genetic services, Poor public education on benefits of genetic services, Low priority in relation to other medical needs of the patient, Long wait time for an appointment)

Did your primary care provider offer any services or resources at this time? How knowledgeable did your primary care physician appear to be regarding the condition?

Did any other specialist offer any services or resources, or help you obtain a genetics referral?

Where do you look to find available genetic services now?

What are the major barriers or challenges you face when accessing genetic services now? (probe for Geography (too far from clinics), Poor or no insurance coverage, Poor provider education on need for genetic services, Poor public education on benefits of genetic services, Low priority in relation to other medical needs of the patient, long wait time for an appointment)

Have you ever participated in a telemedicine genetics visit?

What did you like about it? What did you dislike?

Would you be willing to see your or your child's genetics provider using telemedicine if it meant you could get an earlier appointment?

Can you tell me why telemedicine is not a good option for you or your family?

In your opinion, what are the top two changes you would recommend to facilitate provision of better access to genetic services in your community?

Literature Summary: American Indians

1. Havasupai biologic material collected for diabetes research was used for genetic studies for which specific informed consent was not obtained (schizophrenia, migration, inbreeding)
2. Anonymity not respected at publication of findings
3. Lawsuit filed 2004, well publicized, settled in 2010
4. Based on own and Havasupai experience, Navajo Nation placed moratorium on genetic testing, still in effect

Literature Summary: American Indians

5. Blood and other specimens used for genetic testing are sacred; tampering can cause psychological and/or physical health issues
6. Younger tribal members are more open to benefits of genetic research and testing but still have concerns
7. NCI Genetic Education for Native Americans (GENA) seeks to inform college-age American Indians about genetics
8. Preference for traditional healing over western medicine continues for many tribal members
9. Cultural differences and language make some tribal members hesitate to see western providers
10. Indian Health Service providers turn over frequently in some clinics
10. Logistical barriers such as long distances can limit services

Literature Summary: Hispanic Families

1. Families may be wary of genetic testing because of consent issues
2. Genetic literacy is limited
3. Awareness of genetic services and their uses/benefits may be low
4. Informing influential members of the community (priests, tradespeople, barbers) may be useful
5. Traditional healing practices are affordable and accessible; discussing genetic services with traditional healers may be a good strategy
6. Educating western providers about common illnesses and their remedies in traditional healing may help families access both kinds of services
7. Language barriers, lack of insurance, long distances can limit access to services

8. Cultural differences can be a major barrier to care: familismo, personalismo, respeto, simpatia, confianza
9. Fatalismo (everything is happening as God intended) may shape family's outlook on genetic services (mortality more than morbidity)
10. Promotoras and patient navigators have been shown to increase efficiency of clinical care

Literature Summary: Common Factors

1. Impact of genetic services on the family
2. Ethical issues related to genetic testing
3. Changing standards of informed consent
4. Patient concerns about what happens to their biological samples and genetic information
5. Fear of discrimination based on genetic testing
6. Long clinic wait times and long travel distances to genetics clinics
7. Need for on-site translation services
8. Family cultural preferences

Provider Interview: Tuba City, AZ

1. Tuba City general pediatric clinic provides the only pediatric clinic for approximately 80 miles in any direction
2. There are no genetic specialists at this site (including no adult genetics provider)
3. Nearest genetics clinic is in Phoenix, AZ, about 250 miles away
4. Need is for MD genetics provider rather than genetic counselor
5. High incidence of SCID and Lynch syndrome in the Navajo population
6. Barriers: limited health literacy, long wait times for Phoenix genetics clinic, distance

7. Extensive use of cell phones and Facebook in Navajo population
8. Lay community health workers have limited genetics knowledge and no experience in assisting families with genetic disorders
9. Tuba City clinic has equipment and infrastructure for telegenetics. Major barrier is a clinical geneticist to provide the service.
10. All 8 clinics on reservation lands have similar needs and could benefit from services.

Tuba City UPP Recommendations

1. **Genetics hotline for Tuba City pediatrician, Dr. Steve Holve**
2. **Telegenetics clinic in Tuba City**
3. **Algorithms for initial work-up of referring diagnosis to geneticist (e.g. developmental delay) by primary care provider**
4. **Facebook and other social media outreach to American Indian families**
5. Locate or develop resources for explaining basic genetics for WIC clinics
6. Community Health Worker training
7. Educational resources for families on telemedicine before the first telegenetics visit
8. Explore improvements in overnight mailing of specimens

Tuba City Implementation

1. Telegenetics Clinic and genetics hotline planned for early 2018
2. PCP algorithm on genetic testing for developmental delay in preparation
3. Facebook messaging in preparation for Navajo population (genetic terminology in English and Dine' language with graphics)



Approach to Genetic Diagnosis in Developmental Delay

Initial considerations

- The American Academy of Pediatrics has published their statement on appropriate genetic evaluation of children with developmental delay: see Moeschler et al Pediatrics 2014.
- Much of the following is adapted from that source, with updates based on changing technology for genetic testing
- Reasons to pursue diagnostic genetic testing:
 - A definitive diagnosis (reduced uncertainty, access to support groups, reduction in invasive diagnostic testing)
 - Ability to provide a more detailed prognosis for the child
 - Potential for treatment specific to the diagnosis
 - Determination of recurrence risk for the parents and other family members

● History and physical examination

- Is there a known family history of a specific genetic condition?
- Does the history or physical exam implicate a specific genetic diagnosis based on physical features or characteristic history?
- Is there an aspect of the history that makes a genetic diagnosis less likely (eg extreme prematurity, prenatal exposure to alcohol, history of traumatic brain injury, history of meningitis)?
- If targeted testing is indicated then can contact genetics for advice on the logistics of testing or refer at that time
- If no specific diagnosis is likely then consider the untargeted approach presented below

● MRI of the brain

- Not necessarily indicated in all patients with developmental delay
- Higher yield in the following settings:
 - Epilepsy
 - Macrocephaly
 - Microcephaly
 - Focal neurological findings
 - Developmental regression
- If MRI findings are specific (eg Leigh syndrome) then consider targeted testing for that indication with advice from genetics or refer at that time
- If MRI is not obtained or if findings are normal or nonspecific then consider the untargeted approach presented below

Untargeted approach to genetic testing for developmental delay

- Tier 1
 - Chromosomal microarray
 - Provides copy number of most clinically significant genes (eg deletion, duplication, triplication); can diagnose aneuploidy
 - Roughly 2 week turnaround time
 - Prior authorization should be obtained prior to sending (or use a lab that will complete insurance authorization for you)
 - Informed consent should be obtained prior to testing
 - Fragile X
 - Caused by a trinucleotide repeat
 - X-linked disorder, testing is indicated in both sexes
 - Insurance authorization and informed consent should be obtained prior to testing

- Tier 2
 - Large gene sequencing panel of developmental delay associated genes
 - Several labs will obtain prior authorization for you
 - Informed consent
 - Any time child and parents are tested, there is the possibility to reveal that one of the parents is not the biological parent of the child

- Tier 3
 - Metabolic testing probably has a yield of about 3%, higher in the setting of clear developmental regression
 - See www.treatable-id.org for a testing algorithm and information on diagnoses
 - Informed consent:
 - Algorithm includes:
 - Serum amino acids
 - Serum homocysteine
 - Urine creatine metabolites
 - Urine organic acids
 - Urine purines and pyrimidines
 - Urine oligosaccharides
 - Urine mucopolysaccharides

GLOSSARY FOR BASIC CANCER TERMINOLOGY IN THE NAVAJO LANGUAGE

*A TRANSLATED GUIDE FOR CULTURALLY SENSITIVE
EXPLANATIONS*

*FOR MEDICAL CLINICIANS, EDUCATORS, INTERPRETERS,
RESEARCHERS AND STUDENTS*

Martha A. Austin-Garrison, Lead Translator

Edward R. Garrison, General Editor

DINÉ

Provider Interview, San Antonio, TX

1. Genetics Clinic at San Antonio Children's Hospital sees newborns and pediatric patients; MFM group at San Antonio Children's sees prenatal patients for genetics.
2. Wait time for a genetics clinic appointment is over 6 months. Few PCPs order initial genetic tests because of insurance pre-authorization problems. Genetic counselor often speaks with PCP clinic staff rather than physician about pre-authorization. Education and training of clinic staff might be useful.
3. Barriers: insufficient number of genetics providers; long wait time for insurance companies to authorize genetic tests; reluctance of PCPs and referring hospitals to initiate genetic testing for relatively simple conditions (e.g. Down syndrome)
4. Prenatal genetic services are available by telemedicine in New Braunfels, TX

UPP Recommendations: San Antonio

- 1. PCP education: a) develop algorithms for ordering specific genetic tests; b) develop training modules for PCP staff re' ordering genetic tests and pre-authorization**
- 2. Reach out to referring hospitals re' doing genetic tests on neonates and other in-patients, process of pre-authorization**
3. Consider social media outreach to families
4. Initiate F2F relationship to assist families
5. Locate or develop resources for explaining basic genetics for distribution in WIC clinics
6. Explore cultural competency resources for providers
7. Locate or develop resources on Medicaid and Title V for families

UPP San Antonio Implementation

1. PCP algorithm for genetic testing of development delay in preparation
2. Training module for PCP staff re' pre-authorization of genetic testing planned 2018
2. Outreach to referring hospitals re' genetic testing authorization planned 2018



Provider Interviews: Durango and Grand Junction

1. These clinics (outreach and telegenetics) see general pediatric genetics patients (newborn screening typically seen in Denver first)
2. 9 month waiting list for new genetics patient at these sites
3. Laboratory testing for Medicaid patients is facilitated by PCP ordering through Colorado Children's Hospital, which absorbs costs if Medicaid does not pay
4. Greatest provider need: adult geneticist (current adult genetics provider in Denver has 2 year waiting list)
5. Major barriers: lack of geneticists; poor or no insurance coverage (Tricare does not authorize any genetic testing; private insurance pre-authorization is complicated and time-consuming); poor provider and public education on genetics and genetic testing; long wait time for services; long distance from urban clinics; hospitals unwilling to go through pre-authorization process
6. Solutions: telegenetics clinics; algorithms for genetic testing;

UPP Recommendations: Durango and Grand Junction

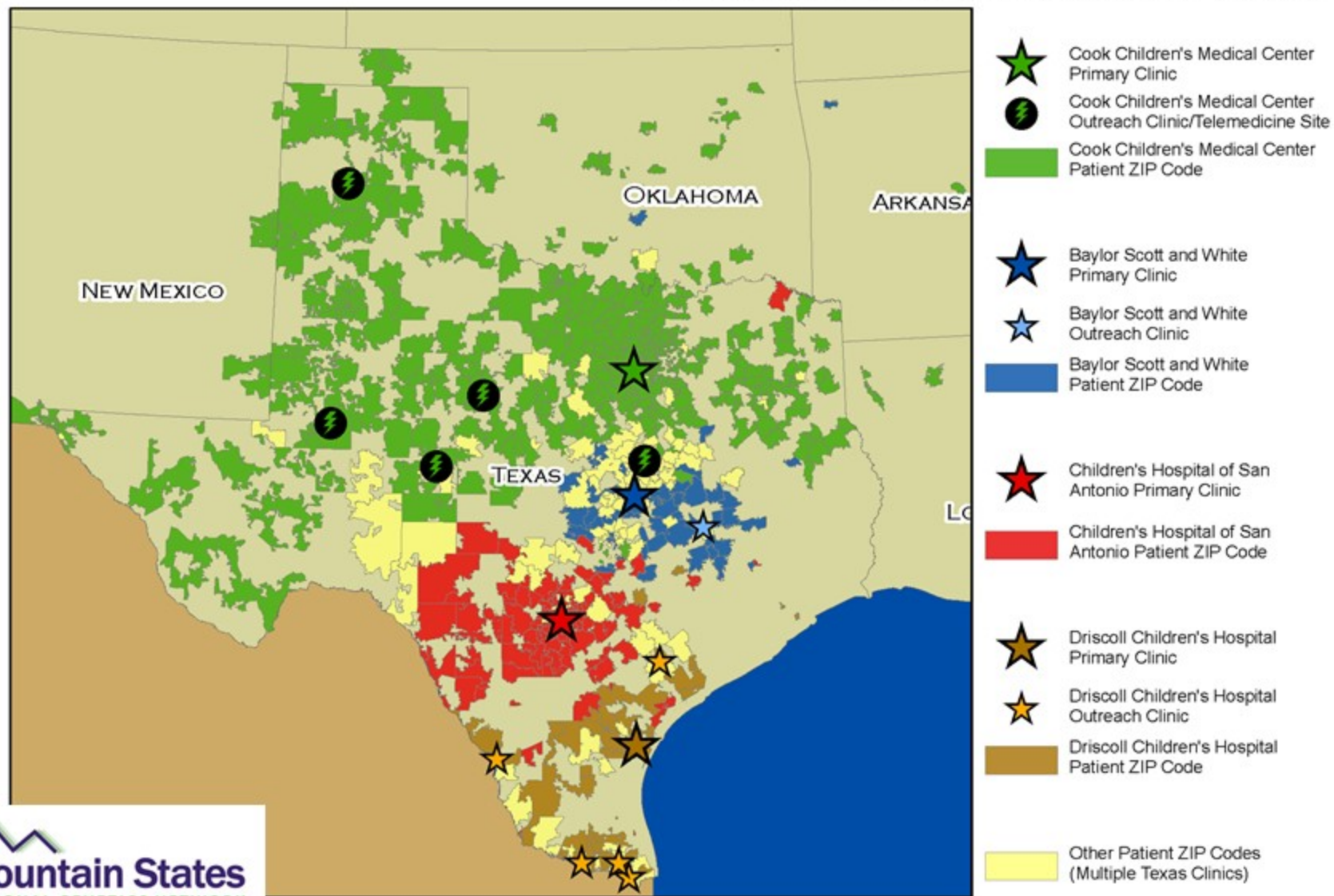
1. **PCP education: develop algorithms for ordering specific genetic tests**
2. Social media outreach to families
3. **Initiate F2F contacts, public health nurses to provide support to families**
4. Locate or develop resources for explaining basic genetics for distribution in WIC clinics
5. **Explore cultural competency resources for providers**
6. **Locate or develop resources on Medicaid and Title V for families**

UPP Implementation: Durango, Grand Junction

1. PCP algorithm for genetic testing for developmental delay in preparation
2. Outreach to public health nurses to provide family support planned 2018
3. Cultural competency resources for genetics providers to be identified (e.g. Hispanic Access Project, Heartland)
4. Identification of Medicaid and Title V Resources in Colorado



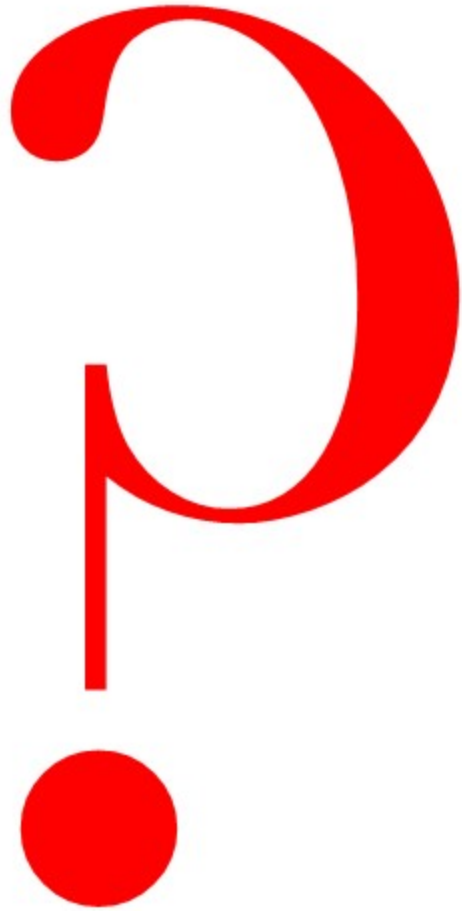
Clinic Locations and Patient ZIP Codes



After UPP

1. Complete implementation and evaluation of UPP in Phase I sites
2. Identify successful strategies to increase access at Phase I sites
3. Provide Phase I data and resources to all state teams
4. Ask state teams to identify areas of underserved populations in each state, review possible strategies for implementation
5. Steering Committee and Management Team to identify next sites for project development and implementation





COMMENTS?
QUESTIONS?