

National Evaluation of the Regional Genetics Collaboratives

Outcome Measures

Evaluation Domain	Outcome Measure	Definition
A1. Improved care coordination for people with heritable disorders	Increase the percentage of states/territories in the region with collaborations facilitated by the regional collaborative between PCPs and specialty (including genetic) providers to improve care coordination for people with heritable disorders	“Collaborations” may include, but are not limited to: learning collaboratives, work groups, practice models and consortia. A collaboration may be at the state/territory level, e.g. between PCPs and specialty providers within a state/territory, or may be at the regional level, provided the collaboration involves both PCPs and specialty providers from the state/territory being counted.
B1. Improved access to genetic services for people with heritable disorders	Increase in the number of genetic services visits provided to people with or at risk for heritable disorders through distance strategies implemented by the regional collaborative.	“Genetic services visit” is defined as “an encounter between a proband/consultand and a genetics provider.” “Distance strategies” may include, but are not limited to: outreach/satellite clinics, formalized long-distance consultation arrangements, telemedicine approaches.
C1. Development of regional/interregional emergency backup	Increase in the percentage of states/territories in the region that have received current materials or other assistance from the RC on developing back-up systems for NBS and genetic services	
D1. Implementation of expanded NBS	Increase in the percentage of states/territories in the region that have evaluated and made recommendations on implementing the ACHDGNC recommended NBS panel	In order to be counted, states/territories can have evaluated and made recommendations and/or have actually implemented the panel.
E1. Improved follow-up of children identified with heritable disorders through NBS	Increase in the number of NBS follow-up specialty visits provided to families through distance strategies implemented by the regional collaborative	“NBS follow-up specialty visit” is defined broadly. These visits can include short-term and/or long-term follow-up. “Distance strategies” may include, but are not limited to: outreach/satellite clinics, formalized long-distance consultation arrangements, telemedicine approaches
E2. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in the region with systems in place to track entry into clinical management for newborns who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening programs	“Entry into clinical management” means that “ a health care provider has accepted responsibility for treatment and/or monitoring of the child”
E3. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in their region with systems in place to track entry into clinical management for newborns who are diagnosed with hearing loss through their State-sponsored newborn hearing screening programs	“Entry into clinical management” means that “a healthcare provider has accepted responsibility for treatment and/or monitoring of the child”

Evaluation Domain	Outcome Measure	Definition
E4. Improved follow-up of children identified with heritable disorders through NBS	Increase in the percentage of states/territories in the region with systems in place to track receipt of clinical services and/or health outcomes for children who are diagnosed with condition(s) mandated by their State-sponsored newborn blood spot screening program and/or with hearing loss through their State-sponsored newborn hearing screening programs	
F1. Improved/expanded education of PCPs about treatment of people with heritable disorders and about clinical genetic resources in region	Increase in the percentage of state/territories in their region whose NBS programs disseminate “just-in-time/point-of-care” information on specific heritable disorders to PCPs	“Just-in-time/point-of-care information” may include ACT sheets, modified ACT sheets or similar information
G1. Improved regional planning around delivery of genetic services to people with heritable disorders	Completion of regional genetic services plan	Plan should at a minimum, include support for optimal diagnosis, genetic counseling, follow-up, and management of people with heritable disorders
G2. Improved regional planning around delivery of genetic services to people with heritable disorders	Annual review and/or update of regional genetic services plan	Plan should at a minimum, include support for optimal diagnosis, genetic counseling, follow-up, and management of people with heritable disorders