HARVARD MEDICAL SCHOOL

Please select the role(s) you perform in your current position as a genetic counselor in the UDN.

Recruitment/application review

- [ ] Assist applicants/referring providers in applying
- [ ] Coordinate collection and organization of medical records and/or additional information from applicants
- [ ] Elicit and construct family histories
- [ ] Review and summarize applicant medical records and/or prior genetic test results
- [ ] Oversee/manage applicant list, physician assignment, and local application review meetings.
- [ ] Oversee/participate in Coordinating Center communications with [prospective] applicants
- [ ] Participate in case review committee (local and UDN-wide) [e.g. presenting, making recommendations for acceptance and/or evaluation strategy]
- [ ] Process applications (data entry) and/or submit accepted cases to Gateway for Case Review Meeting
- [ ] Provide clinical genetic counseling and/or referral letter for [prospective] applicants
- [ ] Oversee/participate in recruitment and outreach activities

Acceptance/consent

- [ ] Communicate acceptance decision to applicants (i.e. composing, sending Accept/Do not accept letters with or without recommendations from review meeting)
- [ ] Communicate acceptance decision to referring providers
- [ ] Discuss genetic testing portion of the UDN consent with participants
- [ ] Discuss entire UDN consent with participants
- [ ] Oversee participant webpages consent
- [ ] Schedule in-person or remote consent discussions with probands and family members

In-person visit

In person visit

- [ ] Participate in development of clinical evaluation plan (e.g. specialist evaluations, clinical tests, etc).
- [ ] Coordinate the logistics and scheduling for the in-person visit and communicate them to the participant/family members
- [ ] Act as participant advocate by accompanying them through most parts of their in-person visit
- [ ] Prepare medical history and prior testing summaries for in-person clinical evaluations
- [ ] Communicate with providers before visit to give them overview of process, to discuss candidate genes, and/or solicit summaries for wrap-up
- [ ] Compose some or all of wrap-up document to be shared with participant and family
- [ ] Meet with participants at the wrap-up meeting

Genetic test/analysis

- [ ] Manage genetic testing strategy (e.g. WES vs. WGS, family members to include, etc.), discuss with clinical team
- [ ] Coordinate some or all of genetic testing for participants and family members (blood draws, sequencing requests in Gateway, billing for sendouts, etc.)
- [ ] Assist in curation and interpretation of raw WES/WGS data
- [ ] Review sequencing report variants (+/- research variants) and assess clinical relevance
- [ ] Communicate genetic results to clinical team, formulate recommendations for follow-up
- [ ] Track progress of each participant through curation pipeline and update clinical team weekly
- [ ] Communicate clinical and/or genetic information to bioinformatics team to improve analysis
- [ ] Submit variants for Sanger validation, track results
- [ ] Coordinate non-UDN reanalysis of sequencing data

Omics, etc. (research and analysis)

- [ ] Manage sendout, progress, and receipt of '-omics' data (RNA, metabolomics, functional studies, etc.)
- [ ] Review and integrate results of '-omics' data (RNA, metabolomics, functional studies, etc.)
- [ ] Lead or participate in curation meetings to discuss updates, plans, and findings from genetic and research studies
- [ ] Initiate participant webpages and track/respond to feedback

- [ ] Work with bioinformaticians and data scientists to develop variant interpretation pipelines and curation tools
- [ ] Study design, data collection/analysis of site- and network-wide studies
- [ ] Enter candidate genes and/or clinical info into case matching databases (e.g. GeneMatcher, KOMP, etc.)
- [ ] Manage communication with research cores

Return of results/closeout

- [ ] Communicate diagnoses to referring providers for transition of care
- [ ] Return genetic results to participants, provide counseling around diagnosis (if appropriate)
- [ ] Document genetic diagnoses in Gateway and other relevant databases for network sharing
- [ ] Write family letter to explain new diagnosis and how to be tested for familial variants if desired/available
- [ ] Return clinical test results to participants (e.g. biochemical, other lab)
- [ ] Compose GC consult and/or clinical genetics note, input into to participant's medical record

Genetic counseling/psychosocial

- [ ] Assist in obtaining appropriate logistical or financial resources (e.g. letter of medical necessity, NORD funds)
- [ ] Identify and/or connect with appropriate support resources (e.g. SWAN, local support groups, diagnosis-specific social media groups) when desired
- [ ] Elicit narrative, participant/family member goals, perform psychosocial assessment, and provide supportive counseling where appropriate
- [ ] Provide anticipatory guidance regarding research process and participants' specific goals
- [ ] Provide psychosocial counseling around diagnoses/results and their implications

Communication with participants

- [ ] Act as main point of contact for participants, families, and/or their advocates regarding all study-related communication
- [ ] Elicit participants' preferences for data sharing post-diagnosis (e.g. inclusion in manuscripts, collaborating with external researchers, etc.)
- [ ] Act as "genetics" point of contact for participant/advocate questions, updates, and needs
- [ ] Participate in or manage PEER group

Communication with external providers

- [ ] Communicate pertinent clinical and/or research information to external providers
- [ ] Manage communications with or inquiries by external experts and researchers regarding candidate diagnoses and/or genes of interest
- [ ] Facilitate transfer of participant samples to outside researchers (e.g. MTAs, shipping logistics, etc.)
- [ ] Educate local providers on how to refer participants to UDN, our process, etc.

Dissemination of knowledge

- [ ] Coordinate local grand rounds/hospital presentations
- [ ] Participate in writing/reviewing manuscripts from UDN team
- [ ] Staff UDN booth at national conferences
- [ ] Present poster and/or platform talks at national conferences
- [ ] Present cases at local/network grand rounds or other meetings
- [ ] Prepare case slides for presentation at internal or external meetings
- [ ] Oversee external data sharing
- [ ] Give informational presentations to various groups about the UDN (internal research groups, GC groups, student interest groups, etc.)

Communication with network/Gateway

- [ ] Act as main contact for UDN CC team & Gateway notifications – distribute to relevant team members
- [ ] Extract pertinent clinical data and enter it into Gateway / PhenoTips
- [ ] Enter appropriate genetic & research requests (Sequencing, MOSC, Metabolomics) into Gateway
- [ ] Enter relatives into Gateway and define whether affected or unaffected
- [ ] Coordinate and return monthly site-ops updates, diagnosis updates
- [ ] Participate in UDN working groups/subcommittees
- [ ] Manage Phenotips feedback from CC and update with new clinical information as needed

- [ ] Manage UDN technology support operations
- [ ] Supervise gathering, documentation, and organization of functional and technical requirements across UDN sites
- [ ] Upload records and wrap up letters into Gateway as needed
- [ ] Review participant webpages

Program development

- [ ] Review and implement improvements to team workflow & research processes
- [ ] Assist in continuing review
- [ ] Elicit feedback from participants/collaborators/local team
- [ ] Monitor & communicate with UDN members progress and milestone progression
- [ ] Participate in development and implementation of site-specific or network studies
- [ ] Create, develop, and revise Network protocols, including central Institutional Review Board (IRB), research, and clinical protocols
- [ ] Manage local site and/or network Manual of Operations
- [ ] Coordinate student volunteers/work-study help
- [ ] Participate in interviewing/hiring of project personnel
- [ ] Lead or participate in grant writing/preparation

Professional development/supervision

- [ ] Supervise research assistants and/or interns
- [ ] Genetic counseling student supervision
- [ ] Participate in informational meetings/shadowing opportunities for prospective GC students or researchers
- [ ] Participate in greater GC community locally
- [ ] Supervise team of study coordinators who schedule and coordinate participants' visits
- [ ] Participate in professional supervision group
- [ ] Genetic counseling student thesis advisor

Miscellaneous

- [ ] Perform “duties as required”
- [ ] Lead biweekly clinical evaluation team (CET) meetings
- [ ] Lead weekly curation team meetings
- [ ] Manage billing needs and/or site budget
- [ ] Serve as central IRB liaison
- [ ] Manage IRB/regulatory documents related to the study
- [ ] Manage social media content relevant to UDN and local-site projects

Name

Please select your UDN site

- ○ Baylor clinical site
- ○ Duke clinical site
- ○ Harvard clinical site
- ○ NIH UDP clinical site
- ○ Stanford clinical site
- ○ UCLA clinical site
- ○ Vanderbilt clinical site
- ○ Coordinating Center
- ○ Baylor sequencing core

How long have you worked with the UDN, in months?

What percentage of your time is allocated to the UDN as defined by your salary?

What percentage of your time is allocated to the UDN in practice?

In a given month, what 5 things (ranked) do you spent the most time on?

Which roles do you get the most fulfillment out of? Please select roles from above survey options.

Comments