Pancreatitis Due to De Novo PRSS1 Pathogenic Mutations
Melissa Samons, MS, CGC; Brissa Martin, MS, CGC; Jing Wang, MD, FACMG, CGMBS

BACKGROUND
- PRSS1-related hereditary pancreatitis (HP) is typically inherited in an autosomal dominant fashion with reduced penetrance.
- In cases without a family history of pancreatitis, the mutation is likely to be identified in one of the patient’s parents.
- De novo pathogenic mutations in PRSS1-related HP have been previously reported; however, the proportion of cases is unknown.

METHODS
- We performed a retrospective review of approximately 9,000 cases received for analysis of the PRSS1 gene between 2005 and June 2017.
- Data from 2 multigene panels (CFTR, PRSS1, SPINK1 +/- CTRC) as well as sequencing of the PRSS1 gene was included in the analysis.
- De novo alterations were confirmed by our laboratory by re-isolation of the specimens with repeat PCR and analysis.
- Familial relationships in Family 1 were verified by analysis of 7-9 different short tandem repeat (STR) loci.

TAKE-HOME POINTS
- PRSS1-related HP due to de novo alterations is rare; we identified this in less than 0.05% of cases (2/9,000 samples).
- Parental testing and genetic counseling should be considered for accurate risk assessment and appropriate clinical follow-up.

REFERENCES