BMP9-Related Telangiectasia Syndrome

Indications for Ordering
Diagnostic test for individuals suspected to have a telangiectasia syndrome

Test Description
Bidirectional sequencing of entire coding region and intron/exon boundaries of BMP9/GDF2 gene

Tests to Consider
Primary test
Telangiectasia Syndrome (BMP9/GDF2) Sequencing 2010015
• Diagnostic test for individuals suspected to have a telangiectasia syndrome
  o In the absence of pathogenic variants in the ACVRL1, ENG, or SMAD4 gene

Related tests
Hereditary Hemorrhagic Telangiectasia (HHT) Panel, Sequencing and Deletion/Duplication 2009337
• Most comprehensive test to diagnosis a telangiectasia/arteriovenous malformation (AVM) disorder

Hereditary Hemorrhagic Telangiectasia (ACVRL1 and ENG) Sequencing and Deletion/Duplication with Reflex to Juvenile Polyposis (SMAD4) Sequencing and Deletion/Duplication 2009008
• Appropriate initial test for individuals with telangiectases clustered on face, hands, and mouth
  o In conjunction with recurrent nosebleeds
  o With or without internal arteriovenous malformations

RASA1-Related Disorders (RASA1) Sequencing and Deletion/Duplication 2007852
• Appropriate test for individuals with cutaneous capillary malformations, with or without telangiectasia and arteriovenous malformations

Familial Mutation, Targeted Sequencing 2001961
• Useful when a familial variant identifiable by sequencing is known

Disease Overview
Prevalence – ~1/5,000-10,000 for hereditary hemorrhagic telangiectasia (HHT)
• BMP9-related disorder – 1-2% of individuals with HHT