NON-SYNDROMIC PORT-WINE STAINS & STURGE-WEBER SYNDROME

Background: Non-syndromic port-wine stains (OMIM 163000) are congenital capillary malformations, often referred to as birthmarks. Port-wine stains have a red, patchy appearance and typically grow as the child grows. Children who are born with a facial port-wine stain have a 6% chance of having Sturge-Weber Syndrome.

Sturge-Weber syndrome (SWS, OMIM 185300) is a sporadic congenital disorder characterized by neurological and cutaneous symptoms. In addition to facial port-wine stains, children with SWS can present with leptomeningeal angiomatosis, capillary malformations of the choroid, seizures, glaucoma, and intellectual disabilities.

Assay: Point mutation analysis in exon 4 of GNAQ (c.548G→A, p.Arg183Gln) in DNA isolated from blood and skin samples by PCR and Next Generation Sequencing on IonTorrent PGM platform

Utility: To identify this somatic mutation in individuals in order to confirm a diagnosis of Sturge-Weber syndrome or non-syndromic port-wine stains.

Sensitivity: When testing the affected tissues (brain and skin) of patients with SWS or non-syndromic port-wine stains, this missense mutation (c.548G→A, p.Arg183Gln) has been reported to be found in 88% of patients (23 of 26) with SWS. In skin samples alone, 100% of participants (8 of 8) were found to have this mutation in skin tissue from port-wine stain skin and 14% of participants (1 of 7) were found to have this mutation in skin tissue that appears normal. Additionally, 92% of patients (12 of 13) with non-syndromic port-wine stains were found to have this mutation in skin tissue from port-wine stain skin.

Turnaround: 4-5 weeks

Fees: $800

2013 CPT codes: 81401x2