PC3. IDENTIFICATION OF GENETIC VARIANTS IN PARRY ROMBERG DISEASE USING WHOLE EXOME SEQUENCING

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PURPOSE: The causative genetic changes underlying hemifacial atrophy (Parry Romberg disease) have yet to be identified. The purpose of this study was to investigate critical genetic loci that may cause phenotypic changes observed in this patient cohort which often require medical and/or surgical intervention.

METHODS: Patients with hemifacial atrophy underwent reconstruction with a free parascapular flap. 1x1 cm skin specimens were obtained from the affected hemiface at the time of free tissue transfer. Skin samples underwent DNA extraction and whole exome sequencing. Candidate genes were filtered using human phenotype ontology (HPO) terms specific to the Parry Romberg phenotype. NxClinical software was used to filter variants based on variant databases and effect predictions.

RESULTS: Twelve skin samples were collected from eight non-related patients. Patient ages ranged from 4 to 39 years (mean 16 years). 434 genes were identified that were significantly (p<0.05) associated with HPO terms linked to the patient’s phenotype. The 1,146 variants in these gene regions were further filtered to identify 1,002 potentially pathogenic variants based on low population frequencies and predicted damaging effects. Single nucleotide variants were most common, followed by deletion and then insertion events. Candidate genes which may be implicated include TCOF1, COL1A1 and COL4A2.

CONCLUSION: This is the first study to use non-related, diseased skin samples to identify candidate genetic variants which may contribute to the Parry Romberg disease phenotype. Further study of these variants may facilitate targeted molecular intervention to prevent atrophic changes and obviate risks associated with current medical and surgical management.

PC4. THE IMPACT OF BREAST CANCER TYPE, STAGING, AND TREATMENT ON FREE FLAP BREAST RECONSTRUCTION COMPLICATIONS

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PURPOSE: The impact of breast cancer staging and treatment on clinical outcomes after autologous free-flap breast reconstruction (ABR) is not well-established. This retrospective review aims to determine if higher breast cancer staging, hormone receptor positivity, and history of venous thromboembolism (VTE) are associated with greater flap vascular complications after ABR.

METHODS: A retrospective review was conducted examining patients who underwent ABR from 2009-2019. Breast cancer staging, types and treatments were collected. Intraoperative and postoperative complications related to vascular compromise, including intraoperative congestion, postoperative take back for flap concern, and flap loss, were analyzed using chi-squared test.

RESULTS: 1,615 patients underwent free-flap ABR during the study period and were included. Patients with estrogen receptor (ER) positive breast cancer have an increased risk of vascular complication compared to ER-negative cancer (6.0% vs. 2.6% for take back, p=0.033; and 13.7% vs. 7.4% for overall vascular compromise, p=0.006). Patients with human epidermal growth factor receptor 2 (HER2) negative
breast cancer were more likely to have vascular compromise than HER2-positive (12.4% vs. 7.4%, p=0.035). Breast cancer staging, progesterone receptor, triple-negative, and neoadjuvant treatment were not associated with ABR vascular complications.

CONCLUSION: We found that ER-positive breast cancer, HER2-negative breast cancer, and history of VTE were independent risk factors for ABR vascular complications. Breast cancer stage and neoadjuvant treatments were not associated with increased free-flap vascular complications after ABR.

PC5. COST ANALYSIS OF MOHS VS TOTAL SURGICAL EXCISION: A RETROSPECTIVE REVIEW

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PURPOSE: Skin cancer risk is elevated in veterans, Caucasians, and males over age 50, who comprise the majority of patients at the Miami VA healthcare system. Treatments include total surgical excision (TSE) with frozen section (FS) or permanent pathology (PS), and Mohs surgery. Our protocol consists of Mohs procedures performed offsite followed by reconstruction at the VA. This retrospective study examines the cost difference between TSE and Mohs surgery.

METHODS: A retrospective chart review was performed of VA patients who underwent TSE or Mohs surgery between 2017 and 2019. Patients under age 18 or those without malignancy on final pathology were excluded. Patients were subdivided into TSE vs Mohs. Cost per OR minute was determined using published data for similar institutions. Pathology costs were estimated using institution specific Medicare data. T-test was performed using SPSS.

RESULTS: Of 130 patients identified, 82 underwent TSE and 48 underwent Mohs with reconstruction. Cost per OR minute for inpatient government owned facilities was $37.94. A flat fee of $1400 for the Mohs surgery was the contracted rate with the offsite institution. Average cost of Mohs surgery with reconstruction was $3534.12. Average cost of TSE with pathology was $2643.85. TSE was significantly more cost efficient than Mohs with reconstruction (p<0.01).

CONCLUSION: At our institution, TSE appears more cost effective than Mohs with subsequent reconstruction. While these are generalized costs, and data specific to our institution, cost efficiency is an important consideration in improving the value of care for VA patients.

PC6. COMPARING GAINS IN FUNCTION AFTER TRADITIONAL TENDON AND NOVEL NERVE TRANSFER FOR PEOPLE WITH CERVICAL-LEVEL SPINAL CORD INJURY

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PURPOSE: Tendon (TT) and nerve transfer (NT) surgery may improve function in people with cervical-level spinal cord injury (SCI)—but which is best? The purpose of this study was to quantify the degree of improvement after TT, NT, and no surgery in the setting of cervical SCI.

METHODS: This prospective multi-center study included adults with mid-level cervical SCI. Health outcomes (ability to accomplish activities of daily living and health status) were assessed using the Spinal Cord Independence Measure III (SCIM) and Short-Form Health Survey (SF-36). Demographic, surgical, and survey data were collected at the