3477

Impaired emotion recognition accuracy after right-hemisphere stroke

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OBJECTIVES/SPECIFIC AIMS: Every year, approximately 800,000 Americans suffer a stroke. Supportive social environments are recognized as an important factor contributing to successful stroke recovery, yet, stroke lesions can affect brain areas important for socioemotional functioning, which could impair a patient's ability to maintain their social relationships. Specifically, emotion recognition, a fundamental socioemotional skill, is predominantly right-lateralized and may be impacted by right-hemisphere stroke. This research tests for emotion recognition impairments after right-hemisphere stroke and examines whether such deficits are associated with worse reported social support. METHODS/STUDY POPULATION: Twenty right-hemisphere stroke patients (9 female, 11 male) and 23 age-matched healthy control subjects (9 female, 14 male) completed laboratory testing including the Geneva Emotion Recognition Test Short. Subjects additionally completed a measure of self-reported social support using the Older Americans Resources and Services questionnaire. Emotion recognition accuracy was calculated using overall accuracy and valence accuracy (i.e. correctly rating a positive emotion as positive). RESULTS/ANTICIPATED RESULTS: Right-hemisphere stroke patients had lower overall emotion recognition accuracy than controls (patients; $M = 37.8\%$, $SD = 18.9\%$. controls; $M = 48.5\%$, $SD = 14.6\%$, $t(41)=2.11$, $p=.041$). Furthermore, patients had significantly lower valence accuracy (patients; $M = 84.5\%$, $SD = 10.7\%$. controls; $M = 90.0\%$, $SD = 5.2\%$, $t(41)=2.19$, $p = .035$), indicating that they more often mistook a positive emotion as a negative emotion, and vice-versa. Finally, within the right-hemisphere patient group, overall emotion recognition accuracy was trending to be positively correlated with self-reported social support ($rho = 0.397$, $p = .083$), suggesting that poor emotion recognition skills may be associated with worse social outcomes in the real-world. DISCUSSION/SIGNIFICANCE OF IMPACT: Our findings indicate that right-hemisphere stroke is associated with impaired emotion recognition. Future research could investigate whether an emotion recognition training may be beneficial for right-hemisphere stroke patient recovery.

3323

Juvenile Polyposis Syndrome Patients Without a Mutation in SMAD4 or BMPR1A: Clinical Presentation and Novel Drivers of Disease

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OBJECTIVES/SPECIFIC AIMS: Juvenile Polyposis Syndrome (JPS) is an inherited cancer predisposition syndrome sometimes attributed to a germline mutation in SMAD4 or BMPR1A. However, many patients meet clinical criteria for JPS without having a pathogenic alteration in either gene. Herein, we perform a cross-sectional analysis of JPS patients at a pediatric and adult tertiary referral center to understand potential differences in the clinical presentation and outcomes of patients with or without a known causative gene mutation. Additionally, we conduct whole exome sequencing (WES) on a subset of the pediatric patients to evaluate for novel genomic drivers of disease. METHODS/STUDY POPULATION: Data were abstracted from medical charts using IRB-approved protocols at the Children's Hospital of Philadelphia (CHOP) and the University of Pennsylvania (Penn). Records were reviewed for patients with a clinical diagnosis of JPS and genetic testing result and seen at either institution in the last 10 years (2008-2018). Patients recruited for sequencing were consented for blood draw through the CHOP IRB protocol, and had whole exome sequencing completed at 70X depth, with data analyzed through institutional pipeline. RESULTS/ANTICIPATED RESULTS: Records were reviewed for 41 patients at CHOP and 19 patients at Penn, for a total of 60 JPS patients. Mean age of CHOP cohort was 11 years: 58.5% male, mean length of follow up 3.9 years. Mean age Penn cohort was 33 years: 47.4% male, mean length of follow up 9.3 years. In the pediatric cohort, 7 patients (17%) had a mutation in BMPR1A (n=6) or SMAD4 (n=1); in the adult cohort, 15 patients (79%) had a mutation in BMPR1A (n=3) or SMAD4 (n=12). The average number of polyps in the pediatric cohort was not significantly higher in patients with a SMAD4 or BMPR1A mutation (9.3 polyps/year of surveillance with a SMAD4 or BMPR1A mutation, vs 5.7 polyps/year; p=0.19). In combined cohort review, all individuals that required gastrectomy and/or colectomy (n=8) as well as all those who developed gastrointestinal cancer (n=3) had a mutation in SMAD4 or BMPR1A. Of the patients who underwent whole exome sequencing (n=13), potential causative germline mutations were identified in four patients (30.8%); all potential drivers identified were within the TNF/BMP pathway. DISCUSSION/SIGNIFICANCE OF IMPACT: This data from a dual-institution review demonstrates that the rate of SMAD4/BMPR1A mutation in JPS is lower in a pediatric cohort compared to an adult cohort. Furthermore, although individuals with JPS may have similar clinical presentations in childhood regardless of whether or not a causative mutation is present, the presence of a mutation in SMAD4 or BMPR1A is associated with a more severe course of disease in adulthood. Further study and a larger cohort will be required to fully validate these findings. Approximately 30% of patients who underwent germline WES had a potential novel driver identified, with further validation underway.

3122

Longitudinal Recovery of Speech Motor Function Following Facial Transplantation

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OBJECTIVES/SPECIFIC AIMS: Using a novel biomechanical-based motor speech assessment alongside commonly used clinically-based motor speech assessments, the goal of this study was to describe longitudinal recovery in speech movements and functional speech in a cohort of 5 patients following facial transplantation. METHODS/STUDY POPULATION: Five participants who had received either full or partial face transplantation were included in this study. Each participant received a unique facial graft from their donor, which included varied amounts of soft tissue, facial musculature, nerve, and bone. Two participants were early in the recovery period and were assessed from zero to 24 months post-transplantation. Three participants were late in the recovery period and were assessed from 36 to 60 months post-transplantation. Each participant completed two data collection sessions and the average time between sessions was 20.4 months. At each session, orofacial movements were recorded using a 3D motion capture system. A 4-sensor head marker

https://doi.org/10.1017/cts.2019.245 Published online by Cambridge University Press