Adult and Pediatric Neurology Case Studies

Chromosome Microarrary Case Examples

Case 1 – Jack is a 4-year-old male with developmental delays who was found on chromosome microarray to have multiple abnormalities. He was referred for genetic counseling to review these results. To aid in the assessment of the clinical significance of these abnormalities, the genetic counselor was able to coordinate for additional studies to be done on Jack and his parents. This additional testing revealed that Jack has a chromosome translocation that was inherited from his father. Through genetic counseling Jack and his family were able to receive a more complete picture of the chromosome abnormalities affecting the family, information was able to be provided regarding the implications for Jack and his family including recurrence risks, and testing was able to be coordinated for other family members.

Case 2 – Elizabeth is a 20-year-old female with a longstanding history of developmental delays and mild intellectual disability who had never undergone genetic testing or evaluation. She was referred for genetic counseling to aid in the coordination of first tier genetic testing – such as a chromosome microarray. This testing revealed that she has a 16p11.2 duplication – a more common chromosome abnormality known to cause intellectual disability and autism. For the first time, the family was able to receive a unifying diagnosis for Elizabeth. She and her mother were able to meet with the genetic counselor to review this diagnosis, implications in respect to Elizabeth's management, and to review recurrence risks.

Case 3 – Taylor is a 7-year-old male with developmental delays. He was referred for genetic counseling due to a variant of uncertain significance seen on chromosome microarray. Specifically he had a deletion on chromosome 5 that overlapped the Cri-du-Chat syndrome region, but was not consistent with the diagnosis. Upon review of Taylor's family history it was determined that his mother was similarly affected. The genetic counselor was able to review the available literature and help coordinate family studies. It was determined that Taylor's deletion involved a critical gene that when involved in the deletion is associated with developmental delays. Other similar cases had been reported, and these patients only had developmental delays and did not have other features seen with those who have larger deletions on chromosome 5. This helped provide guidance to Taylor's developmental pediatrician regarding management. In addition, they were able to provide a diagnosis to Taylor's mother and help her receive services she had not previously qualified.

Neurology examples

Case 1 – Dystonia – Sarah is a 13-year-old female who had a history of possible episodic dystonia. She was referred by her neurologist to help coordinate genetic testing. Through review of Sarah's medical and family history, and in review with her neurologist, the genetic counselor was able to help select the most appropriate gene panel. This involved analysis of a panel of genes associated with various dystonias and ataxia syndromes. Genetic testing revealed that the patient had ATP1A3-related disorder, which is causative of a spectrum of disorders including dystonia, hemiplegia, and ataxia. This diagnosis was then able to be used by Sarah's treating neurologist to help guide her treatment.

Case 2 – Tuberous Sclerosis / Epilepsy – Ryan is a 7-year-old male with complex seizures, who after a detailed physical examination was suspected to have Tuberous Sclerosis. His neurologist coordinate for gene testing for Tuberous Sclerosis which revealed a variant of uncertain significance. Specifically, he was found to have a large duplication involving the TSC2 gene and it was uncertain if this confirmed the diagnosis. He was referred for genetic counseling to review these results and aid in the interpretation of these results along with possible family studies. The genetic counselor was able to meet with Ryan and his family to review the medical and family history in detail, and provide genetic counseling regarding the suspected diagnosis and results. Further testing was able to be coordinated on Ryan and it was determined that his duplication of TSC2 was indeed pathogenic and confirmed his diagnosis of Tuberous Sclerosis.

Case 3 – Epilepsy / Chromosome abnormality – Mary is a 8-year-old female with a history of seizures and developmental delays. Her neurologist was able to coordinate for an epilepsy gene panel to be done, which revealed multiple variants of uncertain significance and indicated several of the genes were deleted (missing a copy). Mary was referred for genetic counseling to help review these results and determine the implications for her and her family. The genetic counselor was able to work together with the neurologist to coordinate further genetic testing, specifically a chromosome microarray. It was determined that the sequence variants detected on Mary's epilepsy gene panel were likely not causative, but that she did have a chromosome abnormality in which epilepsy was a known complication. This information was able to be reviewed in detail with the family, and used to help get familial testing done to determine who else in the family may be affected.

Adult Neurology

Case 1 – CADASIL – John is a 30-year-old male who presented to the hospital with suspected stroke. He underwent further evaluation. Based on his brain MRI which revealed

diffuse white matter lesions and a quick review of the family history, John was suspected to have CADASIL or other related disorder. John was referred for genetic counseling to review the implications of this diagnosis. The genetic counselor was able to meet with John and his family to review the suspected diagnosis, and help coordinate genetic testing. The testing confirmed the diagnosis, and the genetic counselor was able to follow-up with him and review the implications of this diagnosis for him and his family. His neurologist was able to use this information to help guide future treatment and screening options.

Case 2 – HNPP – Lisa is a 67-year-old female who had a longstanding history of peripheral neuropathy. Review of her family history revealed multiple affected generations with similar symptoms including foot drop, weakness of the hands and feet, and transient hand and foot pain that had progressed with age. Lisa was referred for genetic counseling by her neurologist to help coordinate genetic testing. A peripheral neuropathy gene panel was done, and she was confirmed to have the diagnosis of Hereditary Neuropathy with liability to Pressure Palsies (HNPP) – which is caused by a deletion of the PMP22 gene. The genetic counselor was able to review this diagnosis with Lisa and discuss the implications for her other family members.

Questions? Please call us at 844.743.6384.