A PATIENT WITH AUTISTIC SPECTRUM DISORDER AND 17P12 DUPLICATION – LITERATURE REVIEW

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Background There is an increased awareness in the role of microarray analysis in conditions such as ADHD and autism due to arising association of genetic mutations with these diagnoses as risk factors. We have a patient who has been recently diagnosed with autistic spectrum disorder with duplications in the short arm of chromosome 17 with breakpoints within 17p12.

Objectives The purpose is to use the medical literature to search for other cases of 17p12 duplication to see if there is any phenotypic resemblance between the genetic mutation and neuropsychiatric disorders.

Methods We conducted a literature search for other additional cases of 17p duplication as well as specifically looking for 17p12 duplication. We used database engines such as PubMed, National Centre for Biotechnology Information and the Cochrane Library as well as using information from Unique.

Results 17p12 duplication is a rare disorder with approximately 50 people having been diagnosed in the medical literature. With 17p there are four groups that patients tend to fall into depending on specific breakpoints. With 17p12 can have an association with Charcot-Marie-Tooth Type 1a disease if the peripheral myelin protein (PMP22) gene is also included. Duplications with less than 1Mb are strongly associated with autistic spectrum disorder.

Due to small numbers there is very little data and literature regarding 17p duplication in general and even less on 17p12 specifically. Each patient has a unique phenotypic presentation with varying signs and symptoms. This includes behavioural difficulties (such as autistic features and hyperactivity), intellectual disability and unique facial features.

Our patient initially presented with features of learning difficulties, social communication difficulties, sleep disturbances, repetitive behaviours and being investigated for possible dyspraxia. The neonatal period was uneventful and had normal gross motor development. The duplication was approximately 1.3Mb and was stated on the results form as being associated with Charcot-Marie-Tooth Syndrome Type 1a (CMT1A) (CMIM #118220). However, it did not specify as to whether the PMP22 gene was included. CMT1A is a 1.5Mb duplication on 17p12 and our patient’s duplication was measured at 1.3Mb. This was thought to be a co-incidental finding for the referral reason of autistic spectrum disorder. There is no family history of Charcot-Marie-Tooth and our patient has no symptoms of peripheral neuropathy.

Conclusions 17p12 duplication is a rare genetic mutation with very little cases within the medical literature. However, as chromosomal microarray has become increasingly requested for patients with unexplained intellectual disability or autistic spectrum disorders there may well be further individuals diagnosed in the future.