Metabolic investigations were normal and cytogenetic study revealed normal male karyotype. Targeted array-based comparative genomic hybridization revealed a microdeletion of 540kbp from chromosome 7q33 (136,018,399-136,558,387) × 2 region. This deletion was merely encroaching the gene CHRM2 which has a role in central nervous system functioning, although its exact role in autism has not been elucidated so far. Parental analysis revealed a loss of 42kbp from the same 7q33 (136,258,387-136,300,365) × 2 region in father's side. Maternal chromosomal analysis was normal.

The distal region of chromosome 7q is home to many important genes, the deletion/duplication of which has been reported with varying phenotypes. Matsson, et al. [1] reported association of DGKI (diacylglycerol kinase iota) gene at chromosome 7q33 with developmental dyslexia in Finnish and German cohorts. Contactin Associated Protein-like 2 (CNTNAP2), a member of the Neurexin family gene located at 7q34 has been linked strongly with autism [2,3]. There are several other suspicious loci on different chromosomes which are supposed to be linked with autism. Speech and language region which has been most sought to be associated with autism lies at 7q31-33 with FOXP2 and WNT2 genes in region 7q31 being more specific to speech delay and autism, respectively [4,5].

Genetic anticipation is a phenomenon in which symptoms of a disease manifests earlier after passing on to next generations. Small deletion at 7q33 region in father with larger deletion at the same region in son alongwith early presentation of typical ASD features explains the possibility of anticipation in the inheritance of genes related with ASD at 7q33 region. Further clinical validation is important as it may have practical significance on genetic counselling and timing of surveillance initiation. Further research is needed to explore the underlying mechanism of anticipation related with chromosome 7q33 and autism.

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Prescription of Generic Drugs – Is it Really a Smart Initiative?

A generic drug is a drug which has the same constituents, dosage form, strength and quality as the reference/branded drug and is marketed under a non-proprietary name after expiry of the original drug patent [1]. The Government of India recently announced its mandate to stop issuing license for the manufacture or sale of branded drugs in an effort to promote prescription of only generic drugs for patient care, applicable at all government hospitals [2]. It launched the ‘Jan Aushadhi’ campaign for distribution of these generic drugs [3]. The above policy was introduced to curb the presumed malpractice associated with use of branded drugs, wherein the doctors’ prescription may be biased by pharmaceutical companies. Thus, it was anticipated that by prescribing only generic drugs, the malpractice of dispensing costlier medications would be reduced.

However, the following facts need mention to comprehend the present situation. First, the production and availability for most of the generic drugs is limited to few Jan Aushadhi stores, which have insufficient stock of medicines or are non-functional [4]. The only source of medication-provider for the patient is thus the local pharmacy. This leaves the patient to the mercy of the dispenser, who can dispense any brand available for the generic drug prescribed and supposedly make the ‘most suitable’ drug choice for him. Second, many patients
The above cited example is just one of the many instances of drug mal-dispensing that go unnoticed. Therefore, it is essential to develop a fully functioning generic drug production and distribution market before we change to an ‘only generic drug’ policy. Simultaneously, there should be strict monitoring of quality and price of drugs to prevent manufacture of sub-standard and irrationally priced products. The rationale of prescribing a generic drug can only be justified thereafter in patients’ best interests.

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