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A Novel Clinical Challenge: Coexistence of Genetic Disorders in Three Unrelated Cases of Dual Diagnosis

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Introduction

Technical developments in molecular genetics and cytogenetics have made it possible to define a diagnosis for clinical images that are complicated or unusual. In this study, a genetic analysis finds two multimorbidities: one brought on by balletic sequence variations in a gene linked to an autosomal recessive illness and another by either a copy number variant or a chromosomal aneuploidy. We identified three unrelated patients who had these simultaneous conditions that co-occurred by chance: a homozygous variant in the gene known to cause autosomal recessive ciliopathy, down syndrome, and two variants in the gene known to cause me rosin-deficient congenital muscular dystrophy type . When indications and symptoms are inconsistent with the primary diagnosis, it would be hypothesised that the person may be impacted by two relatively common or rare inherited genetic disorders. All of this might have significant effects on how genetic counselling is conducted, how the prognosis is determined, and how the best long-term follow-up is planned.

Description

Medical genetics covers a wide range of topics and is becoming important for the diagnosis of several common and uncommon genetic illnesses. The uniqueness of various genetic illnesses underscores the need of having sufficient clinical expertise as well as how a multidisciplinary approach is crucial for managing and caring for patients. Each patient receives a diagnostic assessment that is customised to their clinical indications and symptoms. This evaluation results in a differential diagnosis, and the proper genetic testing are used to produce an accurate diagnosis. The most significant contributing factor to numerous congenital anomaly/intellectual disability syndromes is complex chromosomal rearrangements. Investigations can offer the typical associated karyotype in the case of suspected genomic disorders by using a chromosomal microarray analysis to find genetic abnormalities. A thorough examination of complicated cases employing [1,2].

When data are inconsistent with the initial diagnosis, it is important to take into account the likelihood of having two relatively common or rare inherited genetic disorders. The clinical care of these individuals is improved by a thorough diagnosis, which also, of course, completely explains their aberrant phenotype. We present the presence of two separate hereditary problems in three unrelated individuals in accordance with the concept of comorbidities, which indicates the coexistence of two or more ailments in the same individual. One person may have many illnesses for one of three primary reasons: chance, selection bias, or one or more types of causal relationship. The investigation showed that the co-occurrence of two genetic illnesses was accidental and unrelated to a cause [3].

With the use of clinical, phenotypic, and blood samples for genetic testing, we assessed three individuals. Physical examination, family history, additional blood samples, and laboratory testing gathered during genetic counselling all formed part of the clinical evaluation. All participants and parents provided written consent. We took a blood sample for genetic testing and looked at the karyotype. According to established methods, a cell culture of peripheral lymphocytes was created, and G-banded karyotyping with a resolution of banding was carried out. Each person's twenty metaphases were examined, a computer-assisted karyotyping system was used to create karyograms. The International Standard of Human Cytogenetic Nomenclature was used to characterise the karyotype an additional CMA inquiry was conducted [4].

With the aid of Assay Kit, the amount of the DNA samples was calculated. Using an Agilent platform (8 60K or 4 180K oligonucleotide array a CMA was carried out on the genomic DNA and examined using Agilent Cytogenetics software (v. Data processing, labelling, and hybridization were done in accordance with the manufacturer's instructions. The Database for Genomic Variants (http://dgv.tcag.ca/dgv/app/home viewed on was used to report benign copy number variations which were deleted from the findings. All patients had their parental analysed using. In order to give a more precise diagnostic and efficient genetic treatment, the second stage of the study employed and was accompanied by clinical consideration and reconsideration [5].

Conclusion

Genetic studies have become crucial in clinical practise as a result of the quick development of genomic technology. Even with complicated phenotypes or the presence of several hereditary diseases, a good clinical evaluation enables the use of an appropriate genetic test to try to pinpoint the source of a problem. Furthermore, precise genetic advice is crucial for controlling and treating patients. In this study, we focused on the coexistence of two genetic disorders that are unrelated to one another. These illnesses were diagnosed by cytogenetic and cytogenetic testing first, and later, an NGS analysis. In three instances, chromosomal aneuploidy and uncommon pathogen etic variations in well-known disease genes coexisted genetically. For instance, the Down syndrome diagnosis was made clinically.

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Conflict of Interest

There are no conflicts of interest by author.

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