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Commentary

Clinical Description of Fragile X Syndrome

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DESCRIPTION

Fragile X Syndrome, the condition was referred to as the Martin-Bell syndrome was described in two different ways, the first of which deals with the clinical description of multiple affected members of a large family, and the second of which interprets the heredity of the ailment. The work is divided into two parts, the first of which deals with the clinical description of multiple affected members of a large family, and the second of which interprets the heredity of the ailment. The clinical description is straightforward and brief The eight males that were tested "Their mental ages range from two to four years, and they have severe dementia.

There are no distinguishing mental or physical characteristics that have been identified that might help to separate the disease that affects this family from other forms of dementia". The sexual development was described as normal, and macroorchidism was not mentioned. Two females were identified as "backward," and were later discovered to be carriers of a complete mutation of the FMRI gene. The lineage was accurately interpreted as X-linked inheritance, allowing for the exceptionality of two transmitting males in whom the mental disability "was sup-pressed by the existence of some regulating factor. The distribution of cases within the family tree, when viewed in retrospect, is completely consistent with Sherman paradox, which describes an increase in the number and proportion of affected individuals in consecutive generations.

The description of a marker X, that is, an X chromosome with a break towards the end of the q arm, subsequently to become known as the fragile site FRAXA, where the causal gene FMRI is located, is the main subject of 'Lub' account of a family with X-linked ID. Because the propositus appeared with a combination of "mental retardation and However, the phenotypic presented is somewhat bland, with only a big maxilla and low-set, huge ears mentioned. Seven members of the Martin-Bell family were retrieved by Richards, Sylvester, and Brooker (1981), and five of them were found to have the fragile X chromosome. The physical properties, such as macroorchidism, the description of a marker X, that is, an X chromosome with a breakage towards the end of the q arm, subsequently to become known as the fragile site FRAXA, where the causal gene FMRI is located, is the main emphasis of the FXS even Lubs' account of a family with X-linked ID [1]. Because the propositus appeared with a combination of "mental retardation and However, the phenotypic presented is somewhat bland, with only a big maxilla and low set, huge ears mentioned. The following description is intended to depict the usual phenotype of a male patient, whose most prevalent physical and behavioral characteristics are provided in the table below.

However, it should be noted that FXS is no exception to the rule that ID and other congenital anomaly syndromes have a wide range of expressivity, making clinical identification challenging at times. Another topic that will be discussed is how the phenotype changes as one gets older.Physical signs are usually less noticeable in infancy [2-4]. However, a missed diagnosis after the age of two years, when developmental milestones are already being missed, should not be justified.A brief description of the clinical presentation of afflicted females will also be provided. The FXS phenotype is produced by inactivation of the FMR1 gene, which results in an expansion of more than 200 units (full mutation) and subsequent methylation of the CGG triplets and CpG island in the promoter region of the gene in more than 95 percent of reported instances.

Point mutations and deletions have been documented seldom, despite the fact that more widespread use of NGS and MLPA techniques may reveal that such alterations are not as uncommon as previously thought. It's possible that the phenotypes aren't always FXS. For example, De Boulle et al. report a case with a significantly more severe and complex clinical picture. Only ID and seizures are shown in 2014 Manifestations in the Physical [5].

The physical phenotype of FXS is primarily displayed in males, and it is usually mild, consisting of small anomalies that can be described, to a significant extent, as due to muscle hypotonia and a connective tissue dysplasia that results in a velvety skin texture. Males with relative macrocephaly, a long and narrow face and a forehead, hypotelorism, a prominent jaw, large and anteverted ears, and a high arched palate are prevalent.

A 3D examination of facial images of patients, both male and female, can provide an accurate and useful description of the craniofacial characteristics of FXS.Spatulate fingers, deep palmar and interphalangeal wrinkles, hyper extensibility of the wrist, and

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interphalangeal folds are all characteristics of the hands. Joints As a result of lax ligaments and muscular hypotonia, droopy shoulders, pectexcavatum, valgism of the elbow and knee, and flat feet are prevalent.The most common symptom is enlarged testes , which can reach a capacity of 50 mL. multiple minor defects the clinical condition is known as syndromal.

Macroorchidism, which affects nearly every adult, is rarely seen in children until puberty. Interstitial edoema, an increased amount of lysosomal inclusions in Sertoli cells, and disruption of spermatid differentiation were found in testicular biopsies under light and electron microscopes. Although evidence is sparse, there is evidence that spermatogenesis is disrupted, resulting in decreased fertility [6].

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