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FA16A

Sociocultural aspects of consanguinity

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Consanguinity is usually defined as the result of a sexual reproduction between two related individuals. It can also refer to populations sharing at least one common ancestor, as those who live within isolates or within communities practicing endogamy. Second or higher order related couples and their offspring represent more than 10% of the current world population. The highest levels of consanguinity are found in the Southern and Eastern shores of the Mediterranean Basin, and the most concerned region extends from the southern shore of the Mediterranean Sea to Southeast Asia through Middle-East, Gulf and India. In Maghreb countries, consanguineous marriages are wide-spread. The rates for this practice vary from 23% in Morocco to 60% in Tunisia, with highest rates being found in rural areas. In Algeria, consanguineous marriages represent more than 38% of all marriages. Large scale migrations from South countries to North countries in the second half of the twentieth century had legal impact on migrants for these specific unions. As a consequence, controversies have been rising in the United States and the United Kingdom especially when a fast decrease of inter-related individuals unions seems unlikely. Consanguinity certainly increases the risk of autosomal recessive pathology, but what about mental pathologies with complex and polygenic heredity? The necessity of an awareness of the genetic risks of consanguinity is as essential in countries where inter-cousin unions are culturally encouraged as among migrant populations in Europe.

Keywords Consanguinity; Genetic; Heredity; Culture

Disclosure of interest The authors declare that they have no competing interest.

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FA16B

Consanguinity and psychosis in Algeria. A family study

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Several studies have affirmed the existence of a strong and complex genetic component in the determination of psychotic disorders. However, the genetic architecture of these disorders remains poorly understood. GWAS studies conducted over the past decade have identified some associations to low effect, and the major part of this heritability remains unexplained, thus calling into question the hypothesis of “common disease – common variant” for model involving a large number of rare variants. Family studies of extended pedigrees selected from geographical isolate can be a powerful approach in identifying rare genetic variants of complex diseases such as psychotic disorders. Here, we studied four multigenerational families in which co-exist psychotic and mood disorders and a high rate of consanguinity, identified in the northwest of Algeria. This case-control study aimed to characterize new rare genetic variants responsible for psychosis. These families have received complete clinical and genealogical investigations, genome wide analysis that were performed in the laboratory of medical genetics in the university hospital of Geneva. A genome wide research CNVs using Agilent Human Genome CGH Microarray Kit 44 K, covering 45 subjects including 20 patients and in a control population of 55 individuals. Three CNVs that had never been reported to date have been identified in one of four families and validated by two techniques. It is the dup 4q26, and 16q23.1 del del21q21. These CNVs are transmitted by either parent line, suggesting a cumulative effect on the risk of psychotic disorders. Further analyzes using pan-genomic linkage analysis using GWAS chip (Illumina Human 660W-Quad v1.0 Breadchip) and complete WES (by GAIIX Illumina/HiSeq 2000) were performed in some related individuals to search other mutations may explain the appearance of the phenotype in this population.

Keywords CNV; Consanguinity; Genetic; Psychotic; GWAS; WES

Disclosure of interest The authors declare that they have no competing interest.

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FA16C

Consanguinité et prédisposition génétique à l'épilepsie

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Le rôle important attribué à la consanguinité dans le développement de maladies à déterminisme génétique est bien documenté ; cependant, son association à l'épilepsie a été suggérée par certaines études et réfutée par d'autres. En Algérie, vu le taux élevé de mariages consanguins (38%), il nous est apparu nécessaire de réaliser une enquête épidémiologique dont l'objectif principal est d'étudier la relation entre la consanguinité et l'épilepsie, non seulement dans la population générale mais aussi parmi des familles algériennes comptant plusieurs membres atteints d'épilepsie.

Méthodes Ont été inclus des épileptiques non apparentés âgés de plus de 16 ans, suivis au service de neurologie du CHU d'Oran et consultant entre octobre 2013 et mars 2014. Des témoins apparus au sexe et à l'âge ont été sélectionnés parmi les patients suivis