Short Communication

CLINICAL INVESTIGATION

Brief note on Corneal ectasia in mothers of children with Down syndrome

Abstract

In this study, corneal results for Keratoconus (KC) and early KC were compared between Mothers With Down Syndrome Children (MDS) and a group of age-matched mothers with normal children (MNC). A clinical symptom and at least one aberrant tomographic or biomechanical criteria were used to diagnose KC. Early KC was characterised as having no clinical signs but at least one aberrant tomographic or biomechanical criteria. The tomographic and biomechanical characteristics of the normal subgroups in each group were compared. The prevalence rates for KC in MDS and MNC were 6.5% and 1.6%, respectively (P=0.047), and 30.9% and 14.3%, respectively (P=0.014). The mean index of height asymmetry, irregularity index, anterior asphericity, pentacam random forest index, corneal stiffness parameters at fi rst applanation, deformation amplitude ra tios, integrated radius-1 mm, highest concavity deflection amplitude, biomechanical corrected IOP, peak distance, and radius (all P 0.05) differed significantly between the two normal subgroups. In this study, MDS was shown to be more likely to have KC as well as thinner, steeper, and softer corneas than MNC. This study's findings underscore the need for more research into the risks of having a child with Down syndrome.

Keywords: Keratoconus • Mothers With Down Syndrome Children (MDS) • Corneal

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Anette Jacobs*

Editorial Office, Journal of Clinical Investigation, London

*Author for correspondence:

clinicalinvest@escienceopen.com

Corneal ectatic

Corneal ectatic diseases can cause vision loss as a result of gradual corneal thinning. Keratoconus is diagnosed, classified, and graded based on clinical indicators, advanced corneal imaging data, and biomechanical features. Although the specific a etiology o f KC i s unknown, past r esearch has shown that it is a complex illness caused by a mix of various variables, including genetics and environment. Because genetic factors play a key role in the development of KC, its prevalence can be linked to other genetic illnesses and syndromes.

Down Syndrome (DS) is a genetic disorder characterised by an extra chromosome 21 (known as trisomy 21) and some degree of intellectual disability. According to earlier data on DS samples, the incidence of KC among these individuals is 0.5%-15% (100 times -300 times greater than the general population). The greater frequency of KC in this cohort might be attributable to the predomi--nance of DS patient's eye rubbing behaviours and other associated genetic collagen-related illnesses. Although there have been numerous research on the frequency of KC in DS patients, there has been no analytical investigation on the relationship between corneal ectasia in moms and DS delivery. This concept was initially

offered by Ambrosio in a case report-3 on a mother with moderate KC who had a DS kid. Given that DS is one of the leading causes of intellectual impairment in the world and can influence many parts of patients' lives, identifying its probable linked components is critical [1].

Because KC and DS are both uncommon disorders, case-control research looking at the relationship between outcome (DS children) and exposure (KC in mother) backward is preferable to long-term follow-up of a large population for a cohort study. This study compared visual and corneal abnormalities suggesting ectasia in moms who had given birth to

Down syndrome children to an age-matched group of mothers who had given birth to normal children [2].

This is the first clinical trial to compare corneal tomography and biomechanical features of MDS and MNC. The findings showed that KC and moderate, fruste (or subclinical) KC were more common in MDS, which also exhibited thinner, steeper, and softer corneas than MNC, even in the absence of definitive KC.

KC is a complex condition caused by a mix of hereditary and environmental factors. The precise reason, however, remains uncertain. Multiple investigations have indicated and verified the importance of genetics in the pathogenesis of KC, and several loci and single nucleotide polymorphisms responsible for KC have been found. Such data suggests that this illness has genetic variability and a complicated aetiology. The superoxide dismutase 1 gene has been studied as a potential candidate gene for KC in various research. This gene encodes a cytoplasmic enzyme that detoxifies superoxide radicals, a kind of reactive oxygen species, lowering cellular oxidative stress. Because increased oxidative stress levels in corneal cells appear to be a risk factor for KC, it has been postulated that a mutation in SOD1 increases oxidative stress and is implicated in the pathophysiology of ectatic diseases. Nonetheless, there is no agreement on the involvement of SOD1 in KC, and there is conflicting data. SOD1 is the sole KC candidate gene found on chromosome 21, and it may explain the link between KC and DS in some way. DS arises in 95 percent of instances when a gamete with two chromosomes 21 (owing to nondisjunction of chromosome 21 pairs in meiosis) crosses with a normal gamete to form a zygote cell with three chromosomes 21 (trisomy 21). Numerous research has looked at and established the link between DS and KC. Although blepharitic eye rubbing is one of the most common causes of KC in DS patients, genetic flaws in the structure and composition of these individuals' corneas can also be a source of ectatic diseases. Thus, the genes responsible for corneal biomechanical defects, refractive errors, and the development of ectatic illnesses such as KC in DS patients may be found on chromosome 21. However, it is probable that age-related abnormalities in these specific genes render women more prone to chromosome 21 nondisjunction in meiosis, increasing the likelihood of DS newborns. SOD1 is one of these genes that has been related to KC; there may be more genes that have yet to be found. In this regard, the findings of this study show that aberrant KC indices are more prevalent in MDS than in MNC, although these abnormalities are more indicative of moderate KC [2-5].

The study has limitations, but it opens up a new research field for the correlations between KC and the chance of a mother having a child with DS. Following the initial anecdotal account, this is the first clinical investigation with large enough sample numbers to discover statistically significant differences between MDS and MNC. The subjects were drawn from various sources in Iran, making the findings more generalizable. The individuals in this study were not genetically tested for KC-related genes (e.g., SOD1). Due to the rarity of KC and DS, the study design was confined to a correlation study; otherwise, we would have been able to conduct their relationship with greater power. To test this idea, we conducted a case-control research with a reduced power instead of recruiting a cohort of KC women throughout their reproductive age and monitoring pregnancy outcomes in terms of DS birth. Despite the decreased power, there were statistically and clinically significant differences between MDS and MNC.

The current study discovered that MDS are more likely to have KC as well as thinner, steeper, and softer corneas than MNC. Such findings suggest the need for more research but should not be used to determine the risk of having a kid with DS at this time. Indeed, multimodal corneal imaging, including corneal tomography and biomechanical evaluations, should be used in bigger retrospective research in order to establish a significant prospective study.

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