Please find below the results of your literature search request.

If you would like the full text of any of the abstracts included, or would like a further search completed on this topic, please let us know.

We’d appreciate feedback on your satisfaction with this literature search. Please visit http://www.hello.nhs.uk/literature_search_feedback.asp and complete the form.

Thank you

**Literature search results**

<table>
<thead>
<tr>
<th>Search completed for:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Search required by:</td>
<td>ASAP</td>
</tr>
<tr>
<td>Search completed on:</td>
<td>29th May 2015</td>
</tr>
<tr>
<td>Search completed by:</td>
<td>Lesley Firth</td>
</tr>
</tbody>
</table>

**Search details**

Siblings with cystic fibrosis (clinical management issues, infection control, psychological issues, social issues, genetic counselling)

**Resources searched**

NICE Evidence; TRIP Database; Cochrane Library; CINAHL; MEDLINE; PsychINFO; Google Scholar

**Database search terms:** "cystic fibrosis" OR CF, (paediatric* OR pediatric* OR child* OR "young person" OR "young people" OR adolescence*), (sibling* OR sister* OR brother* OR twin*), (family OR familial), "infection control", (psychology* OR social* OR psychosocial), ("genetic counselling" OR "genetic counselling"), (manage* OR treat* OR support*)

**Evidence / Google Scholar search string(s):** "cystic fibrosis" (sibling OR twin)

**Guidelines and Policy**

**Archives of Disease in Childhood Education and Practice**

*Diagnosis and management of cystic fibrosis*, 2005

Makes no specific mention to managing siblings with CF

**International Society for Pediatric and Adolescent Diabetes**


From p. 4
A genetic association between CF and type 2 diabetes is suggested by the increased prevalence of type 2 diabetes in monozygotic vs. dizygotic twins with CF.

**Journal of Pediatrics**

Cystic Fibrosis Foundation evidence-based guidelines for management of infants with cystic fibrosis, 2009

Not open access – please request if needed.

**Evidence Reviews**

Nothing found

**Published Research – Databases**

**Title:** Several siblings with Cystic Fibrosis as a risk factor for poor outcome.

**Citation:** Respiratory medicine, Jan 2015, vol. 109, no. 1, p. 74-78 (January 2015)

**Author(s):** Lavie, Moran, Shemer, Ofer, Sarouk, Iflat, Bar Aluma, Bat el, Dagan, Adi, Efrati, Ori, Vilozni, Daphna

**Abstract:** Occurrence of Cystic Fibrosis (CF) in more than one member in a family is not uncommon. The aim of our study was to assess the influence of multiple siblings with CF on disease expression and outcome. Study group consisted of 2-siblings (2-sibs, n = 42) or 3/4 siblings (3/4-sibs, n = 22) with CF in one family. Each sibling was matched by age, mutation, and gender to a single CF patient. 3/4-sibs subgroup compared to singles showed a lower mean FEV1 with a faster decline rate (58.4 ± 27.5 vs. 72.7 ± 25.4 and -5 ± 6.4 vs. -1.7 ± 2.8 %predicted decline/year respectively, p

**Source:** Medline

**Full text:** Available EBSCOhost at Respiratory Medicine

**Title:** The microbiome in pediatric cystic fibrosis patients: the role of shared environment suggests a window of intervention.

**Citation:** Microbiome, Jan 2014, vol. 2, p. 14. (2014)

**Author(s):** Hampton, Thomas H, Green, Deanna M, Cutting, Garry R, Morrison, Hilary G, Sogin, Mitchell L, Gifford, Alex H, Stanton, Bruce A, O'Toole, George A

**Abstract:** Cystic fibrosis (CF) is caused by mutations in the CFTR gene that predispose the airway to infection. Chronic infection by pathogens such as Pseudomonas aeruginosa leads to inflammation that gradually degrades lung function, resulting in morbidity and early mortality. In a previous study of CF monozygotic twins, we demonstrate that genetic modifiers significantly affect the establishment of persistent P. aeruginosa colonization in CF. Recognizing that bacteria other than P. aeruginosa contribute to the CF microbiome and associated pathology, we used deep sequencing of sputum from pediatric monozygotic twins and nontwin siblings with CF to characterize pediatric bacterial communities and the role that genetics plays in their evolution. We found that the microbial communities in sputum from pediatric patients living together were much more alike than those from pediatric individuals living apart, regardless of whether samples were taken from monozygous twins or from nontwin CF siblings living together, which we used as a proxy for dizygous twins. In contrast, adult communities were comparatively monolithic and much less diverse than the microbiome of pediatric patients. Taken together, these data and other recent studies suggest that as patients age, the CF microbiome becomes less diverse, more refractory to treatment and dominated by mucoid P. aeruginosa, as well as being associated with accelerated pulmonary decline. Our studies show that the microbiome of pediatric patients is susceptible to environmental influences,
suggesting that interventions to preserve the community structure found in young CF patients might be possible, perhaps slowing disease progression.

Source: Medline
Full text: Available National Library of Medicine at Microbiome

Title: Facial palsy and idiopathic intracranial hypertension in twins with cystic fibrosis and hypovitaminosis A.
Citation: Pediatric neurology, Feb 2011, vol. 44, no. 2, p. 150-152 (February 2011)
Author(s): Obeid, Makram, Price, Jason, Sun, Linus, Scantlebury, Morris H, Overby, Philip, Sidhu, Reet, Chiriboga, Claudia A, Quittell, Lynne M
Abstract: Facial nerve palsies are uncommon in infants. We report on 10-week-old monozygotic twins, diagnosed with cystic fibrosis by newborn screening, who developed facial palsy and increased intracranial pressure. Cranial imaging and cerebrospinal fluid analysis produced normal results. Levels of serum vitamin A were below normal range. Low levels of vitamin A are associated with facial nerve paralysis, and are at least partly implicated in the development of increased intracranial pressure in infants with cystic fibrosis.

Source: Medline

Title: Genetic counselling issues in cystic fibrosis.
Citation: Paediatric respiratory reviews, Jun 2010, vol. 11, no. 2, p. 75-79 (June 2010)
Author(s): Culling, Bronwyn, Ogle, Robert
Abstract: Cystic fibrosis is a chronic condition for which genetic testing offers much for the individuals affected in terms of an early diagnosis and offers timely additional information for families with regard to family planning and prenatal testing. Genetic counselling encompasses a range of clinical issues for families and forms a complementary resource for clinicians caring for people with cystic fibrosis. This review will discuss the range of genetic information readily available to patients and families through genetic counselling.

Source: Medline

Title: Long-term effects of birth order and age at diagnosis in cystic fibrosis: a sibling cohort study.
Citation: Pediatric pulmonology, Jun 2010, vol. 45, no. 6, p. 601-607 (June 2010)
Author(s): Slieker, M G, van den Berg, J M W, Kouwenberg, J, van Berkhout, F, Teding, Heijerman, H G M, van der Ent, C K
Abstract: Siblings with cystic fibrosis (CF) share many genetic and environmental factors but may present different phenotypes. Younger sibs are mostly earlier diagnosed with CF than their older sibs, but might be at risk for an earlier colonization with Pseudomonas aeruginosa (PA) than their older counterparts due to cross-infection within families. To analyze the effects of birth order and age at diagnosis on lung function, PA colonization, nutritional status, and survival during the first two decades of life in siblings with CF. A retrospective cohort study of 52 sibling pairs was performed in two Dutch CF centers. Data were analyzed both cross-sectionally and longitudinally using Kaplan-Meier curves and modified log-rank tests. Median age at diagnosis was significantly higher in the older sib compared with the younger sib (3.0 and 0.2 years, respectively, P

Source: Medline

Title: The importance of sweat testing for older siblings of patients with cystic fibrosis identified by newborn screening.
Citation: The Journal of pediatrics, Dec 2009, vol. 155, no. 6, p. 928 (December 2009)
Author(s): Munck, Anne, Houssin, Elise, Roussey, Michel
Abstract: We report cystic fibrosis (CF) care center instructions for sweat testing in older siblings after implementation of the French nationwide newborn screening
program, and we evaluate the incidence of unrecognized CF. Nearly 9% of families with an infant screened for CF were unaware of an affected older sibling. We strongly recommend sweat testing for all first-degree older children.

Source: Medline

Title: A family with atypical cystic fibrosis: brother and sister with heterozygosity for both G542X and R117H.

Citation: Pediatric and developmental pathology : the official journal of the Society for Pediatric Pathology and the Paediatric Pathology Society, May 2008, vol. 11, no. 3, p. 213-219, 1093-5266 (2008 May-Jun)

Author(s): Farez-Vidal, M Esther, Gómez-Llorente, M Amelia, Gómez-Llorente, Carolina, Blanco, Sonia, Casas-Maldonado, Francisco, Campoy, Cristina, Gómez-Capilla, José Antonio

Abstract: Clinical manifestations of cystic fibrosis (CF) are variable. Genetic and environmental factors that determine whether an individual will develop associated complications are still under investigation. The present study reports the genetic analysis of a family with different clinical forms of CF and addresses the difficulty of CF diagnosis in an individual with mutant alleles G542X and R117H because of the variable phenotype associated with R117H mutation. Both children in this family were heterozygous for G542X/R117H with the same thymine sequence (7T/9T) in intron 8 of CF transmembrane conductance regulator. The girl was diagnosed with CF, whereas the boy was diagnosed with azoospermia as the sole clinical manifestation. The possible implication of the hemochromatosis gene as a CF modifier locus was analyzed because the 2 children had the same genotype. No genetic differences were detected between brother and sister that explained the different clinical manifestations of CF.

Source: Medline

Full text: Available EBSCOhost at Pediatric and Developmental Pathology

Title: Cystic fibrosis presenting as new onset diabetes mellitus in adolescent twins.


Author(s): Starkman, Harold, Das, Shilpi

Abstract: We describe identical adolescent twin girls who presented with symptoms consistent with type 1 diabetes. Medical work up for evaluation of gastrointestinal symptoms led to a diagnosis of cystic fibrosis (CF) in both. These cases suggest that diabetes can be a presenting symptom of CF in the absence of pulmonary symptomatology.

Source: Medline

Full text: Available EBSCOhost at Pediatric Diabetes

Title: Familial concordance of phenotype and microbial variation among siblings with CF.

Citation: Pediatric pulmonology, Oct 2004, vol. 38, no. 4, p. 292-297, 8755-6863 (October 2004)

Author(s): Picard, Elie, Aviram, Micha, Yahav, Yaakov, Rivlin, Joseph, Blau, Hanna, Bentur, Lea, Avital, Avraham, Villa, Yael, Schwartz, Shepard, Kerem, Batsheva, Kerem, Eitan

Abstract: The clinical spectrum of cystic fibrosis (CF) is influenced by the cystic fibrosis transmembrane conductance regulator (CFTR) genotype. However, variable courses of the disease were demonstrated among patients with identical genotypes. Since siblings share identical CFTR mutations and environmental factors, they can serve as a model to assess the effect of modifier genes on disease expression, and also to evaluate cross-infection. The aim of this study was to compare disease expression among siblings with CF. All sibling pairs treated at 7 CF centers in Israel were included in the study. Data were collected from patients’ medical charts. Fifty families with at least 2 siblings were identified. As
expected, the second-born sibling was diagnosed at an earlier age compared to the first-born. The mode of CF presentation at diagnosis showed significant familial concordance. In the families where the first sibling presented with gastrointestinal manifestations, 79% of the second siblings also presented with gastrointestinal manifestations. When gastrointestinal manifestations were absent in the first sibling, only 12% of the second siblings presented with gastrointestinal manifestations (P < 0.0001). Likewise, when the first sibling presented with respiratory symptoms, 60% of the second siblings presented with the similar symptoms. However, when the first sibling presented without respiratory symptoms, only 12% of the second siblings presented with respiratory symptoms (P < 0.001). Meconium ileus (MI) was present in 20 patients (21%). In 10 families where the first-born sibling had MI, 8 (80%) of the subsequent siblings had MI. On the other hand, in the 39 families where the first-born sibling did not have MI, only 2 (5%) subsequent siblings had MI (P < 0.001). Pancreatic insufficiency (PI) also had high familial concordance (P < 0.0001). Percentile growth for weights and heights and lung function (FVC, FEV(1), and FEF(25-75)) at ages 7 and 10 years were similar between siblings. P. aeruginosa grew from sputum in 89% of our study patients. When P. aeruginosa was isolated from the first-born patient, 91% of the second siblings were also positive for P. aeruginosa, whereas when the initial sibling was not a carrier of P. aeruginosa, only 50% of subsequent siblings were positive (P < 0.0001). This familial concordance was not observed for S. aureus. By contrast, the age of first isolation of P. aeruginosa and S. aureus was significantly earlier in the second sibling than in the first for the two bacteria: 10.3 +/- 5.1 vs. 7.3 +/- 5.2 years (P < 0.05) for P. aeruginosa, and 11.5 +/- 5.4 years vs. 6.8 +/- 5.1 years (P < 0.05) for S. aureus. CF siblings tend to share similar phenotypes that are not mutation-dependent. The lack of variability between siblings in mode of initial CF presentation, rates of MI, pulmonary function, and nutritional status supports the role of modifier genes in the determination of these factors.

Source: Medline

Title: Disease severity in siblings with cystic fibrosis.
Citation: Pediatric pulmonology, May 2004, vol. 37, no. 5, p. 407-412, 8755-6863 (May 2004)

Author(s): Katz, Sherri L, Strug, Lisa J, Coates, Allan L, Corey, Mary

Abstract: Since cross-infection occurs between cystic fibrosis (CF) siblings, we hypothesized that subsequent siblings may acquire respiratory pathogens at an earlier age and have a more severe course of pulmonary disease. We studied a retrospective cohort of 31 sibling pairs from the CF database at the Hospital for Sick Children. Kaplan-Meier curves and modified log-rank tests were used to test sibling differences in age of acquisition of Pseudomonas aeruginosa (PA), Staphylococcus aureus (SA), or any positive culture. Differences in disease severity outcomes were explored. Older siblings were more likely to have both SA and any CF pathogen first isolated from respiratory culture at an older age than younger siblings (P = 0.0050 and P = 0.0008, respectively, by modified log-rank tests). However, more of the older siblings were positive on first culture at time of diagnosis, introducing an age-of-diagnosis bias. Hospitalization rates, courses of oral antibiotics, FEV(1) % predicted, and weight and height measurements were not better in the older children. No differences in clinical parameters were found between older and younger siblings. The apparent finding of younger age at first isolation of pathogens from respiratory cultures in younger siblings is likely because many older siblings were already infected with these organisms at time of diagnosis. Copyright 2004 Wiely-Liss, Inc.

Source: Medline

Title: Disclosing to parents newborn carrier status identified by routine blood spot screening.
Newborn blood spot screening programmes are designed to detect serious conditions affecting individuals, where early treatment can improve health. It is suggested that screening can improve the experience of diagnosis for parents. For example, without newborn screening, when a child with cystic fibrosis becomes symptomatic a period of uncertainty can arise prior to diagnosis. These potential advantages of screening need to be weighed against potential disadvantages of screening at individual and population levels. Some newborn screening programmes inadvertently identify newborn infants who, although not affected by the condition, carry a gene for it and can pass on that gene to their children; these are ‘genetic carriers’. Knowledge of newborn carrier status can lead to: testing of parents and family members, and concern about possible affected future siblings should both parents be identified as carriers; the possibility of such testing revealing the putative father is not the biological father; concern about the child’s future reproductive choices; and unjustified anxiety about the health of the carrier newborn. There is an urgent need to develop clear guidance as to how to respond, with advances in technology fueling the expansion of newborn blood spot screening and raised expectations of informed consent and disclosing test results. Depending on the condition for which screening is offered, options include: employing tests that do not identify carrier status, if available; identifying acceptable ways of disclosing carrier status; or identifying acceptable ways of not disclosing carrier status. These options are illustrated by screening programmes for sickle cell disorders and cystic fibrosis. Currently, there are no screening tests available for sickle cell disorders that do not identify carrier status. For cystic fibrosis, the policy choice is between an extended period of testing, and a screening result that is available sooner for most newborns, but inadvertently identifies carrier babies. The aim of this review was to assess the impact of disclosing to parents newborn carrier status inadvertently identified by routine newborn blood spot screening. We searched for reports addressing disclosing newborn carrier status to parents following newborn screening for sickle cell disorders and cystic fibrosis in: commercially available electronic databases (October 2002), specialist registers, online journals, online abstracts and conference abstracts. We also scanned the reference lists of included papers. Studies addressing the impact of disclosing carrier status using a soundly controlled trial or randomised controlled trial. Two researchers independently scanned titles and abstracts for relevance using the pre-specified inclusion criteria. Full reports of selected citations were then located and screened again for relevance by two researchers independently. At each stage, results were compared and discrepancies resolved by discussion. We found no controlled trials about disclosing carrier status. There is a need to develop and evaluate the effects of interventions to support the disclosure of carrier status to parents following newborn screening.

Source: Medline

Title: Categories of deltaF508 homozygous cystic fibrosis twin and sibling pairs with distinct phenotypic characteristics.

Citation: Twin research : the official journal of the International Society for Twin Studies, Dec 2000, vol. 3, no. 4, p. 277-293, 1369-0523 (December 2000)

Author(s): Mekus, F, Ballmann, M, Bronsveld, I, Bijman, J, Veeze, H, Tümmler, B

Abstract: Cystic fibrosis (CF), the most common severe autosomal recessive trait among Caucasians, is caused by molecular lesions in the cystic fibrosis transmembrane conductance regulator gene (CFTR). The course of the multi-organ disease CF is highly variable, suggesting the influence of environmental factors and/or modulating genes other than CFTR on the disease phenotype. To evaluate the cause of CF disease variability, the European CF Twin and Sibling
Study collected data on two clinical parameters most sensitive for the course and prognosis of CF, ie weight predicted for height (wfh)\% (representative for the nutritional status) and FEVPerc (representative for the pulmonary status) for a cohort of 277 sibling pairs, 12 pairs of dizygous twins and 29 pairs of monozygous twins. Of these 318 CF twin and sib pairs, 114 were reported to be homozygous for the most frequent CF disease-causing lesion, deltaF508. Intra-pair discordance was assessed by the intra-pair differences with wfh\% and FEVPerc and by DELTA, a composite parameter defined by linear combination of wfh\% and FEVPerc in order to describe discordance with respect to the overall disease severity. Monozygous twins had a significantly lower DELTA than dizygous twins (P = 0.05) indicating that CF disease severity is modulated by an inherited component in addition to the CFTR gene itself. Extreme phenotypes are considered to be more informative for the analysis of any quantitative trait. Thus, we aimed to quantify disease severity and intra-pair discordance in order to select pairs with the extreme phenotypes DIS (discordant patient pairs), CON+ (concordant and mildly affected patient pairs) and CON- (concordant and severely affected patient pairs). The algorithm reliably discriminated between pairs DIS, CON+ and CON- among the cohort of deltaF508 homozygotes. The selected pairs from these categories demonstrated non-overlapping properties for wfh\%, FEVPerc and the intra-pair difference of both parameters.

Source: Medline

Title: Residual chloride secretion in intestinal tissue of deltaF508 homozygous twins and siblings with cystic fibrosis. The European CF Twin and Sibling Study Consortium.

Citation: Gastroenterology, Jul 2000, vol. 119, no. 1, p. 32-40, 0016-5085 (July 2000)


Abstract: Cholinergic stimulation of chloride secretion is impaired in the intestines of patients with cystic fibrosis (CF). However, intestinal chloride secretion has been observed in patients with mild CF mutations. The aim of this study was to investigate residual Cl(-) secretion in the intestine of DeltaF508 homozygous CF patients, and examine the contribution of cystic fibrosis transmembrane conductance regulator (CFTR) and alternative Cl(-) conductances. Twins and siblings with identical CFTR genotypes were investigated to determine the impact of factors other than CFTR on chloride secretion. Chloride secretion in rectal tissue was investigated by applying Ca(2+) and adenosine 3',5'-cyclic monophosphate (cAMP)-linked agonists before and after the inhibition of alternative Cl(-) conductances with 4,4'-diisothiocyanostilbene-2, 2'-disulfonic acid (DIDS). cAMP-mediated Cl(-) secretion was observed in 73% of patients, and 20% showed DIDS-sensitive Ca(2+)-activated Cl(-) secretion. This DIDS-sensitive alternative chloride conductance was seen only in CF patients who also responded to cAMP agonists. Chloride secretion was more concordant within monozygous twins than within dizygous pairs. These results suggest the presence of CFTR-mediated Cl(-) secretion in a subgroup of patients, implying that a portion of deltaF508 CFTR can be processed in vivo and function as a chloride channel in the apical membrane of intestinal cells. Moreover, a considerable number of deltaF508 homozygous patients express chloride conductances other than CFTR in their intestinal epithelia.

Source: Medline

Full text: Available the ULHT Library and Knowledge Services' eJournal collection at Gastroenterology

Title: Confidential inquiry into families with two siblings with cystic fibrosis.

Citation: Archives of disease in childhood, Dec 1997, vol. 77, no. 6, p. 501-503 (December 1997)
Author(s): Lane, B, Williamson, P, Dodge, J A, Harris, H, Super, M, Harris, R

Abstract: To audit the care that had been provided to couples before the birth of a child with cystic fibrosis where a sibling had been previously diagnosed. Retrospective review of case notes. Families where at least one affected child had been born between 1 January 1991 and 30 June 1995 and the diagnosis in the first child was made before the second affected pregnancy reached 20 weeks. The combination of information on these families with data from the prenatal diagnosis register allowed the reconstruction of a cohort of pregnancies in women with a previous affected child. Forty six eligible families with a second affected child were identified. Details from the paediatrician who had diagnosed the first affected child were obtained in 43 cases: all 43 couples were offered genetic counselling, but where provided by a paediatrician this was difficult to assess as no couple was sent a summary letter. Details were obtained from the obstetrician in the subsequent affected pregnancy in 42 cases: prenatal diagnosis was not offered in 10 (24%), offered and declined in 24 (57%), offered and accepted but termination declined in eight (19%). In the overall cohort of at risk pregnancies, the estimated rate of prenatal diagnosis offer was 97%, prenatal diagnosis uptake 86%, false negative prenatal diagnosis rate 0%, and uptake of termination 95%. (1) Parental choice was an important determinant of second affected births. (2) Despite widespread availability, prenatal diagnosis was not offered in an estimated 3% of at risk pregnancies. (3) There were shortcomings in counselling documentation, in particular failure to send a summary letter to counselled couples.

Source: Medline
Full text: Available EBSCOhost at Archives of Disease in Childhood

Title: Exchange of Pseudomonas aeruginosa strains among cystic fibrosis siblings.
Citation: Research in microbiology, Jun 1997, vol. 148, no. 5, p. 447-454, 0923-2508 (June 1997)
Author(s): Renders, N H, Sijmons, M A, van Belkum, A, Overbeek, S E, Mouton, J W, Verbrugh, H A

Abstract: The molecular epidemiology of Pseudomonas aeruginosa infection in cystic fibrosis (CF) siblings was analysed by DNA fingerprinting using arbitrary primed polymerase chain reaction. A total of 306 strains collected from six pairs of siblings over a period of 20-126 months (median 64) was studied. Fifty-four different P. aeruginosa genotypes were recognized. Two out of six pairs of siblings were ultimately colonized by identical strains, and it was shown that a single P. aeruginosa clone can persist in an individual patient for over ten years. No overlap in P. aeruginosa genotypes was encountered between families, whereas in all families at least transient cross-colonization with the same genotype was observed. This finding demonstrates that P. aeruginosa cross-infection or acquisition of the same strain from an identical environmental source exists within the family situation, but does not always result in a long-term colonization by identical genotypes in all family members suffering from CF.

Source: Medline

Title: Perception of carrier status by cystic fibrosis siblings.
Author(s): Fanos, J H, Johnson, J P

Abstract: Now that the cystic fibrosis (CF) gene has been identified, direct genetic testing for this disorder is available. The current lack of precision has generated a controversy concerning whether population screening is advisable. However, there is general agreement that testing for CF carriers should be offered to CF-affected families. The purpose of this study was to explore levels of understanding and feelings about carrier status and genetics of CF in affected families. Fifty-four adult CF siblings and their 30 spouses drawn from Children's Hospital, Oakland, and Children's Hospital, Boston, were interviewed, and transcripts were coded on
various categories. The relationship between birth order and beliefs about carrier status was significant, with last-born siblings more likely to believe they are not carriers. Higher sibling resentment was found to be significantly related to willingness to abort an affected fetus, to more guilt, and to assumption of carrier status. Thirty percent of siblings believe that carrier status implies health difficulties. Increased feelings of guilt were significantly related to the belief that carrier status implies health problems and to the wish to be a carrier. Interestingly, beliefs regarding carrier status and the wish to be a carrier are not influenced by educational or income level. Some siblings have had their child tested for carrier status and others are planning to do so before the child reaches the age of 18 years. Perception of carrier status is strongly influenced by psychological factors.

(ABSTRACT TRUNCATED AT 250 WORDS)

Source: Medline

Full text: Available National Library of Medicine at American Journal of Human Genetics

Google Scholar

From the 1st fifty results:

Nothing additional found