Pediatric Rheumatology

Poster presentation

Clinical phenotype and CARD15 gene mutation with Blau Syndrome in Chinese children and their parents

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Background

Blau Syndrome characterized by granulomatous polyarthritis, uveitis and rash with a typical onset before 5 years. We summarized the article to find the clinical features of our patients and analyze CARD15 gene mutation of the patients and their parents.

Materials and methods

Studied on clinic and basis aspect of cases of Blau Syndrome in Beijing Children's Hospital from the year of 2006 to 2007.

Results

8 patients were diagnosed. The onset age was from 1 month to 5 years. Three of them were misdiagnosed as JIA and Takayasu's arteritis respectively. One case had family history. All patients has had typical rash, joints problem, bilateral pan-uveitis. Two had hearing lose, four had Takayasu's arteritis with hypertension, and two of them had renal artery stenosis with severe hypertension and aortitis. Histologically, there was synovial and dermis proliferation with non-caseating giant cell granulomas in all of the patients. We analyzed 6 patients and their parents' NOD2/CARD15 gene. We have found six mutations in them. R334W and R334Q were reported previously abroad, E383D, R471C and R587C are new mutations. In the treatment, all of them received NSAIDS, steroid treatment, one of them also with TNF blockers. All of them were efficiency.

Conclusion

Blau syndrome is a rare auto-inflammatory disease. We diagnosed 8 patients in Chinese Children. That indicate Blau syndrome also can involve Chinese population. They had CARD15 gene mutation and some of the mutations are special changes in Chinese population.