Pediatric Rheumatology

Poster presentation

Prospective validation of the diagnostic score for molecular analysis of hereditary autoinflammatory syndromes in Italian children with periodic fever

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Objective

Aim of the study was to verify in a prospective study the sensitivity and specificity of a recently elaborated diagnostic score for the prediction of the presence of mutation of genes associated with periodic fever [1].

Patients and methods

Detailed clinical information of 100 Italian patients with a clinical history of periodic fever was collected since June 2007. For each patient the Diagnostic score (www.printo.it/periodicfever) was calculated. According to previous experiences a cut-off > 1.32 was chosen to define those patients at high risk to carry relevant mutations. All patients were screened for mutations of *MVK*, *TNFRSF1A* and *MEFV* genes.

Results

Ten patients displayed relevant (homozygous or compound heterozygous) mutations for MVK and MEFV genes. No structural mutations of TNFRSF1A gene were found. 10 patients dysplayed low-penetrance mutations of the TNFRSF1A gene (R92Q) or a single mutation of the MEFV gene. 80 patients were negative to all the three genes.

The Diagnostic score revealed high sensitivity (90%) and specificity (65%) in discriminating positive and negative patients. The regression tree analysis [1] was able to provide the correct identification of the affected gene in 7 out of the 9 positive identified by the diagnostic score.

Conclusion

This study confirm the validity of the Diagnostic score as a useful tool for the identification of children at higher risk to carry relevant mutations of genes associated with periodic fever.

References

1. Gattorno M, *et al.*: **A diagnostic score for molecular analysis of hereditary autoinflammatory syndromes with periodic fever in children.** *Arthritis Rheum* 2008, **58:**1823-1832.