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Note	In relazione con <a href="http://eprints.uniss.it/11602/">http://eprints.uniss.it/11602/</a>
Sommario	<p>Mutations in several genes are associated with epilepsy (e.g. SCN1A, MECP2, ARX). Identifying genetic causes in epileptic syndromes is crucial to avoid a complex diagnostic work up, to provide genetic counseling, to start a tailored treatment in some cases and to avoid drugs potentially worsening seizures in others.</p> <p>Next Generation Sequencing (NGS) technologies allow analyzing a large number of genes in a single experiment, shortening the time to reach a definite diagnosis, and saving costs.</p> <p>Aim of this research was to identify gene variants underlying epilepsies with a challenging etiological classification. DNA from 81 pediatric epileptic patients was analyzed with a gene panel set up by child epileptologists, neurophysiologists and geneticists. This included 55 genes, later extended to 91, associated or not to intellectual disability, additional neurological signs, and complex malformations.</p> <p>In 14 patients pathogenic mutations were individuated, with an overall mutational frequency of 17,2% (14/81). 90,5% of patients had previously undergone unrevealing cytogenetic or single-gene analyses, thus our population was highly selected at the time NGS was performed.</p> <p>It is essential to underline that NGS must not be considered a screening examination, and that it requires a multidisciplinary approach in patients' selection, and results interpretation.</p>

Localizzazioni e accesso

[http://memoria.depositolegale.it/\\*/http://eprints.uniss.it/11602/1/SALIS\\_B\\_Next\\_Generation\\_Sequencing\\_Epilepsy.pdf](http://memoria.depositolegale.it/*/http://eprints.uniss.it/11602/1/SALIS_B_Next_Generation_Sequencing_Epilepsy.pdf)

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