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# The impact of the IKBKG gene on the appearance of the corpus callosum abnormalities in incontinentia pigmenti

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Incontinentia pigmenti (IP) is a rare X-linked genodermatosis with an estimated prevalence of 1.2/100.000 (Orphanet report series, 2020). It appears almost exclusively in females and is usually lethal in males (Landy and Donnai, 1993). It is caused by a mutation of the IKBKG gene localized on the X chromosome.

The most prominent clinical manifestations of IP are skin changes, representing major IP diagnostic criteria (Minic et al, 2014, Clin Gen). Dental, ocular and central nervous system (CNS) anomalies are considered as minor criteria (Minic et al, 2014, Clin Gen).

CNS anomalies usually occur from the neonatal period and represent the most important threat to normal life of patients with IP. One of the most frequent CNS abnormalities found in IP were corpus callosum (CC) abnormalities.

Knowing the frequency of CC anomalies in IP, as well as the fact that the most frequent causes of ACC are gene mutations, the connection between their occurrence and gene mutations in IP patients should be investigated.

The aim of this study was to determine the presence of CNS abnormalities, especially CC anomalies in IP patients, as well as their relationship with the IKBKG gene mutations, possible presence of other gene mutations, and the X-chromosome inactivation pattern.

For this purpose, genetic analyses of the IKBKG gene and the X-chromosome inactivation, as well as Magnetic Resonance Imaging (MRI), Next Genome Sequencing (NGS) and Whole Exome Sequencing (WES) analyses were performed on a group of seven patients with a clinically confirmed diagnosis of IP, according to the updated IP diagnostic criteria.

The simultaneous presence of IKBKG mutation and CC abnormalities, and the absence of other mutations indicates that IKBKG may be the cause of CC abnormalities and should be included in the list of genes responsible for CC abnormalities.

## Biography

Snežana Minić graduated from the Faculty of Medicine at the University of Belgrade and obtained MD in 1986, and a PhD in 2001 on Incontinentia pigmenti clinical aspects, ultrastructural and cytogenetic analyses. Since 1988, she has been working at the University Clinic of Dermatovenerology, Clinical Center of Serbia and Faculty of Medicine, University of Belgrade both as a practitioner and a professor. She is the Associate professor of Dermatovenerology, chief of the Polyclinic's department at the Clinics of Dermatovenerology, Clinical Center of Serbia and Faculty of Medicine in Belgrade.