

že být vztah mezi genotypem a skutečným fenotypem klinicky závažný.

Metodologický pokrok v oblasti NGS a sekvenování exomu tyto limity postupně překonává. Nové technologie umožňují de-

tekce vzácných variant, CNV i přímé fázování haplotypů, které konvenčním metodám chybí. Předpokladem jejich smysluplného zavedení do rutinní praxe však není jen technologická připravenost laboratoří, ale i edukace lékařů a sys-

témová integrace farmakogenetických dat do elektronické zdravotní dokumentace. Výsledek farmakogenetických testů je tedy třeba vnímat nikoli jako definitivní diagnózu, ale jako jeden ze vstupních parametrů klinického rozhodování.

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