P-502 New method for the molecular genetic diagnosis of heritable disorders of connective tissue using next-generation sequencing (NGS).
A A Pushkov, A G Nikitin, A V Pakhomov, N V Jourkova, K V Savostyanov, N D Vashakmadze, L S Namazova-Baranova

P-503 Urinary pyridinoline cross-links in osteogenesis imperfecta
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P-504 The extension of molecular genetic diagnostics of congenital adrenal hyperplasia in the Czech Republic
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P-505 Two cases of pxoprolinuria: 5-oxoprolinase deficiency?
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P-506 Peripheral blood smear and/or bone marrow analysis are simple tools to conduct the diagnosis in inborn errors of metabolism
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P-507 LPIN1 mutations cause severe rhabdomyolysis in childhood - the Austrian experience
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P-509 Metabolic Diet App Suite: Digital medicine to support families with inborn errors of metabolism
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P-510 Atypical clinical presentation of familial hypomagnesemia with hypercalciuria and nephrocalcinosis: report of two Russian siblings
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P-511 Neopterin antioxidant effects in glial cells *K Ghisoni, L Barbeito, A Latini*

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A-001 Case of combination of cystic fibrosis with metabolic disorders of fatty acids and sulfur containing amino acids
A A Yanovska, Y B Grechanina, E Y Grechanina, E P Zdubskaya

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L E Bernstein, C E Burns

A-003 A systematic review (SR) and meta-analysis (MA) to assess blood phenylalanine (Phe) levels in adults with phenylketonuria (PKU)
D A Bilder, J K Noel, E R Baker, W Irish, B J Winslow, R Jain, Y Chen, M J Merilainen, S Prasad

A-004 A systematic review (SR) and meta-analysis (MA) to assess the prevalence of neuropsychiatric symptoms in adults with phenylketonuria (PKU)
D A Bilder, J K Noel, E Baker, W Irish, B J Winslow, R Jain, Y Chen, M J Merilainen, S Prasad

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A-005 Profiling of genetic variants in the folate-mediated one-carbon metabolism (FOCM) pathway as risk factors for hyperhomocysteinemia in a North Indian cohort
A Lomash, S Kumar, S Pandey, A Singh, S K Polipalli, S Kapoor

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A-006 Early diagnosis of Grawitz tumor due to alkaptonuria follow up
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