



DGAP112: 46,XX,t(3;12)(q13.2;q14)

Revised: 46,XX,t(3;12)(q11.2;q14.1),del(12)(q14q14)

Microcephaly, growth retardation, developmental delay, facial dysmorphism and café-au-lait spots are present in this female. In addition, the 2nd toes overlap the 3rd bilaterally, and she has small labia majora, extra creases on the right hand, wider thumbs and halluces, advanced bone age and secondary craniosynostosis. FISH analysis of the 12q14.1 breakpoint revealed a submicroscopic interstitial deletion of 500-700 kb. The deletion contains a single gene, *SLC16A7*, a proton-linked monocarboxylate transporter that catalyzes the rapid transport of many monocarboxylates such as lactate, pyruvate, branched chain oxo-acids derived from leucine, valine and isoleucine, and the ketone bodies acetoacetate, beta-hydroxybutyrate and acetate across the plasma membrane. Interestingly, FISH mapping of the 3q11.2 breakpoint identified a split clone, AC117474, which overlaps with *PROS1*, an anticoagulant plasma protein. As a cofactor to activated protein C in the degradation of coagulation factors Va and VIIIa, *PROS1* functions to prevent coagulation and stimulate fibrinolysis. Defects in *PROS1* are the cause of protein S deficiency, and are associated with an increased risk to develop thrombotic disease.

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