SHWACHMAN DIAMOND SYNDROME MIMICS DISORDER OF GALACTOSE METABOLISM

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BACKGROUND

Shwachman Diamond syndrome (SDS) is a rare autosomal recessive disorder characterized by exocrine pancreatic insufficiency, bone marrow dysfunction and skeletal abnormalities. Massive galactosuria usually occurs in disorders of galactose metabolism.

CASE REPORT

Male patient born from G7P6 at 37 weeks, birth weight 2290g, height 47 cm, Apgar score 8/10. Family history: the first child – severe mental retardation, fifth – Down syndrome (Fig.1).

Soon after birth the child presented failure to thrive, anemia, visually big abdomen, liver +1cm, *S. aureus* sepsis. Urinary screening for IEM showed massive galactosuria. Breast feeding was stopped immediately. Within one month
on galactose restricted diet, there was no noticeable improvement – still failure to thrive, anemia, although no signs
of steatorrhea. GALT and galactose metabolites analyses were negative. Noticeably decreased serum elastase level was detected, due to pancreatic insufficiency and pancytopenia SDS was suspected,
later the diagnosis was confirmed in Verona, Italy, by E. Nicolis, by finding mutations 183-184TA>CT/258+2T>C in *SBDS* gene (Fig.2).

Fig.1 **Patient's family tree**

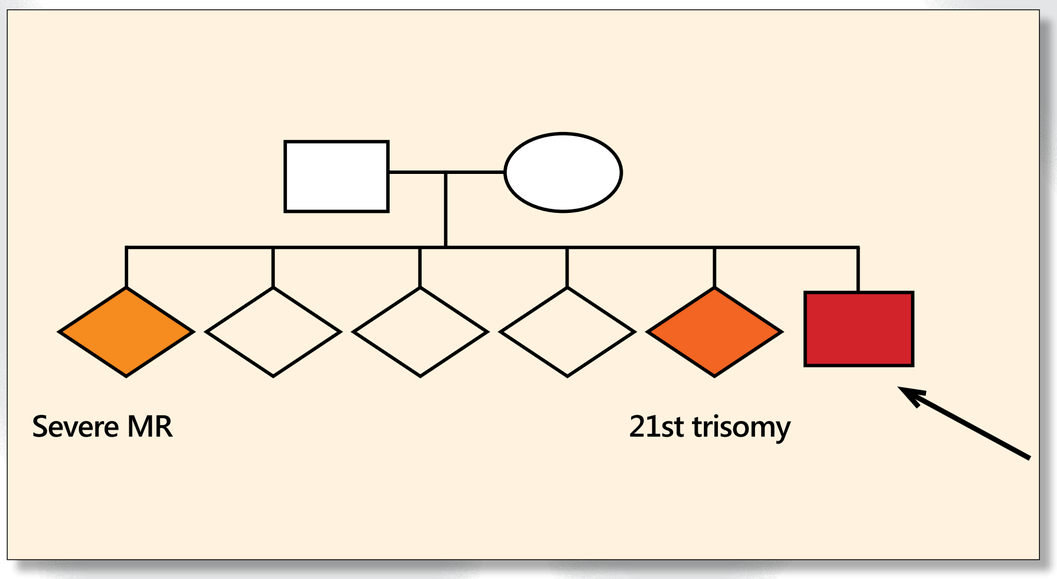
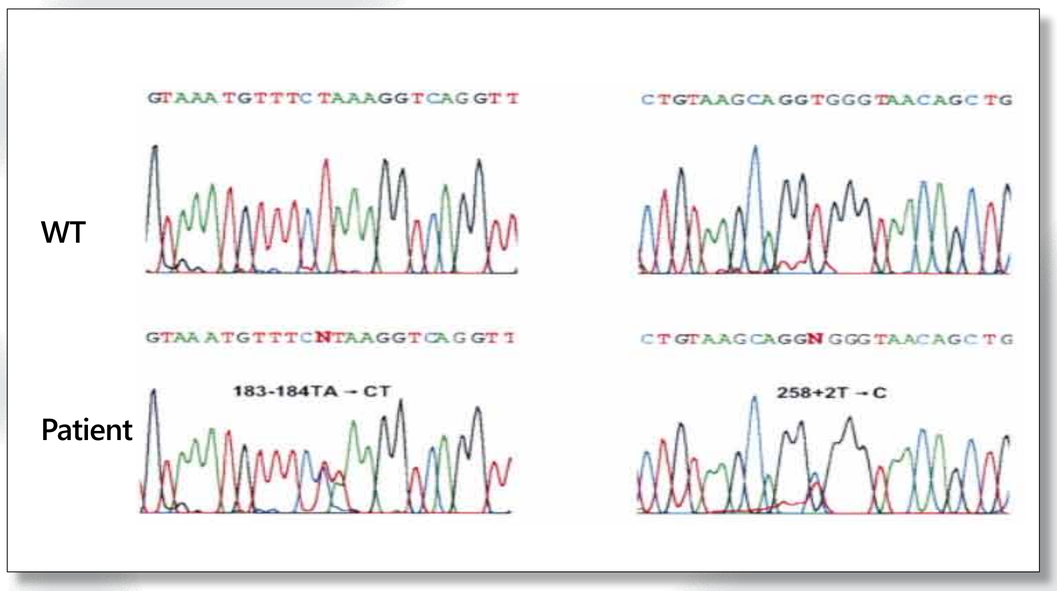


Fig.2 **Mutation analysis**



CONCLUSIONS

According to literature galactosuria is a characteristic sign of SDS, but usually not as noticeable as in our case, so SDS must be considered as differential diagnosis. This case demonstrates a clinical overlap between symptoms of diseases of completely different origin.

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