as a third choice TKI. However, due to the deep neutropenia of the patient, it was possible to continue with a dose of 10 mg/m^2 from the 2nd week. With this dose, the neutrophil is around 0.8-1 $\times$ 10^3/μL. Under ponatinib treatment, BCR-ABL copy number was 6.6% IS at 1 month, 0.8% IS at 3 months, 0.09% at 5 months, and 0.05% at 6 months. No significant side effects were observed except neutropenia. **Conclusion:** There is no approved treatment in pediatric CML cases where the second choice TKI fails and there is no donor for transplantation. FDA approval for ponatinib in adult patients was obtained in December 2020. Ponatinib is a natural or mutant pan-BCR-ABL mutation inhibitor. It also inhibits VEGFR, FGFR,PDGFR,EPH and SRC kinases as well as KIT,RET,TIE2 and FLT3. The use of ponatinib should be evaluated by monitoring side effects/tolerance in pediatric cases where there is no other treatment option, and there is a need for studies on this subject.

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INHERITED BONE MARROW FAILURE DISEASES

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INVESTIGATION OF SALIVARY miR-9, miR-34a ve miR-196a LEVELS IN FANCONI ANEMIA AND ORAL SQUAMOUS CELL CARCINOMA PATIENTS

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Objective: Fanconi anemia (FA) is a rare bone marrow deficiency syndrome due to the DNA repair gene mutations, and Oral Squamous Cell Carcinoma (OSCC) is seen more frequently in FA patients than in the general population. The dysregulation of PI3K and Wnt signaling has been implicated in OSCC pathogenesis and abnormal expressions of miRNAs (a class of non-coding small regulatory RNAs) associated with these signaling pathways has been reported in OSHK patients. Salivary miRNAs are valuable biomarker candidates for OSCC development and prognosis. In this study, salivary levels of miR-9, miR-34a and miR-196a miRNAs related to PI3K and Wnt signaling pathways were examined in OSCC and FA patients and compared with the healthy control group. **Methodology:** Saliva samples were collected from 89 subjects including 25 OSCC patients, 24 FA patients and 40 healthy controls. Total RNA was isolated using Quick-RNA Miniprep Kit (Zymo Research) due to the kit instructions. cDNA was generated with miRCURY LNA miRNA PCR Assay (Qiagen, Hilden, Germany) and Quantitative real-time PCR was performed with miRCURY LNA SYBR Green PCR Kit (Qiagen, Hilden, Germany) according to the manufacturer's protocol. For the normalization of the expression levels of each miRNA, the mean expression U6 SnRNA was used as reference. The ΔΔCt value and the normalized miR-9, miR-34a and miR-196a salivary levels were calculated with Livak Method. **Results:** Our results showed that miR-9 and miR-34a levels in OSCC patients were significantly lower compared to healthy control groups (p= 0,01 and p= 0,012), and there was no significant difference in miR-196a levels (p> 0,05). In FA patients, miR-9 and miR-34 levels were lower than in control groups, likewise the OSCC patients (p =0,017 and p =0,014). There was no significant difference between miR-9, miR-34a, and miR-196a levels of FA patients and OSCC patients (p >0.05). **Conclusion:** According to our results, low levels of miR-9 and miR-34a in saliva are biomarker candidates that may be important for OSCC development. In FA patients, close follow-up of the levels of miR-9 and miR-34 would be appropriate considering OSCC development. Further studies are needed to confirm the potential of miR-9 and miR-34a as biomarkers for OSCC.

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A NOVEL MISSENSE MUTATION OUTSIDE DNAJ DOMAIN OF DNAJC21 IS ASSOCIATED WITH SHWACHMAN-DIAMOND SYNDROME

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Shwachman-Diamond Syndrome (SDS) and related bone marrow failure disorders are characterized by early onset pancytopenia with a hypocellular bone marrow, short stature, and pancreatic insufficiency, along with an increased risk for myeloid malignancies. Recently, several cases with an SDS-like syndrome have been reported to harbor mutations in the DNAJ domain of *DNAJC21*. Here, we report an intriguing case