History
10 year old female with Fanconi anemia and headache.

Diagnosis
Fanconi Anemia

Additional Clinical
Bone marrow transplantation several years ago.

Discussion
Fanconi anaemia is an autosomal recessive disease characterised by congenital abnormalities, defective hematopoiesis, and a high risk of developing acute myeloid leukemia and certain solid tumors. Underlying defect is related to chromosomal fragility and defective DNA repair. Brain calcifications are seen in about 1% of CT examinations and often associated with hypoparathyroidism, pseudohypoparathyroidism, trisomy 21, mitochondrial defects, neurodegenerative disease, some tumors, infections, or bone marrow failure syndromes (such as Fanconi anemia, Revesz syndrome and other myelodysplastic diseases). A suggested explanation of intracranial calcifications in bone marrow failure syndromes is bleeding with subsequent dystrophic calcification related to altered blood cell morphology (inducing endothelial damage) and abnormal blood cell number (predisposing to bleeding).

Findings
CT-Multiple intracranial calcifications mainly junctional but also in deep gray and white matter tracts.

Reference
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