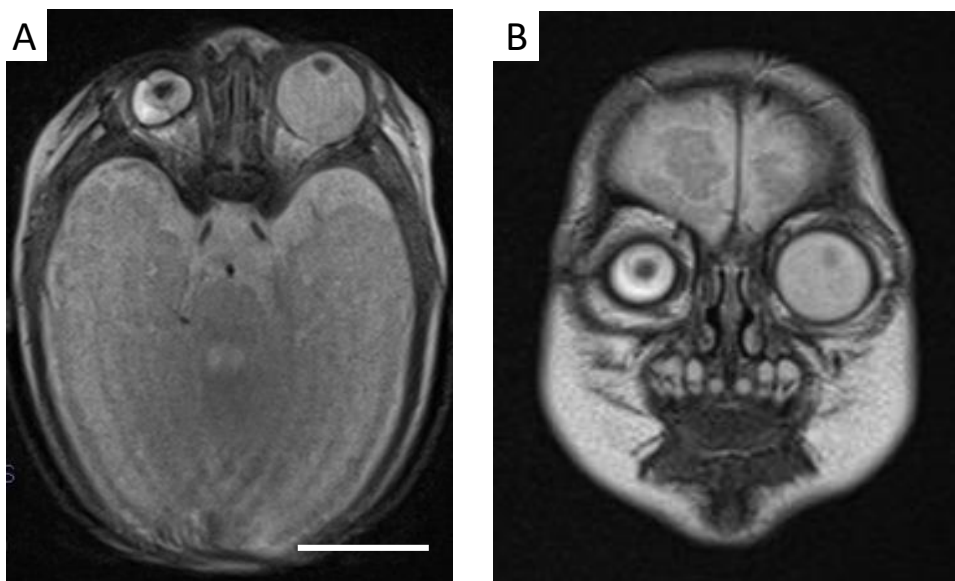
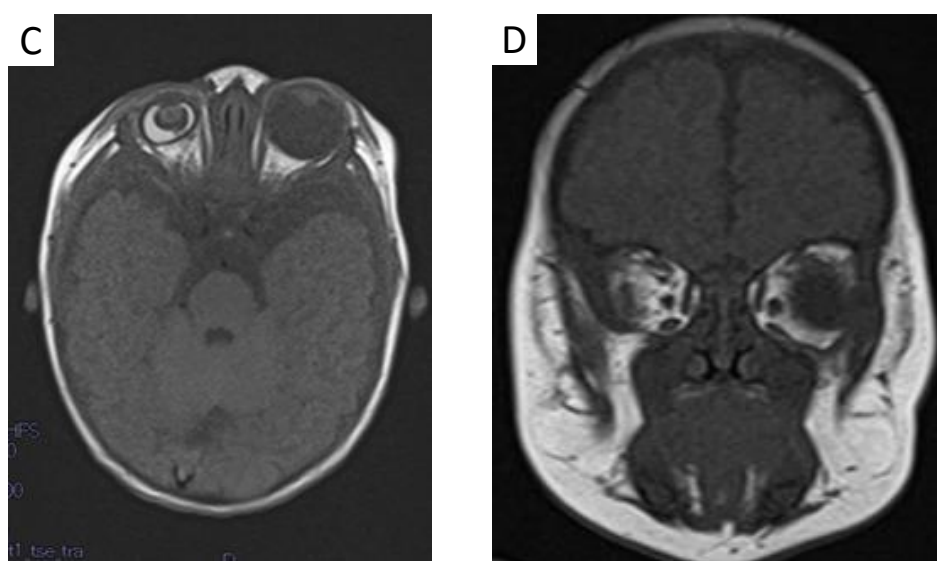


Additional file 2: Clinical phenotype (1)

T2-wighted



T1-wighted



Additional file 2 MRI features of the orbita and brain

T2-weighted image (**A** axial, **B** coronal) and T1-weighted image (**C** axial, **D** coronal) are shown. The right eye ball was remarkably hypoplastic with lens dislocation and thickening of the eye globe wall. The size and organization of left eye appeared grossly normal. The lens was positioned to the anterior chambers but deviated toward upward (*arrowhead*) Optic nerves appeared normal bilaterally (*arrows*). Bar indicate 3cm.