Title: Segmental agenesis of the corpus callosum with pituitary hypoplasia
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Abstract: Corpus callosum dysgenesis is found in 1:4000 individuals. Despite aberrant variations not being particularly rare, current research still aims at reconciling a mechanism for embryologic callosal development. It was once theorized that the corpus callosum develops linearly in a rostro-caudal direction from the genu to splenium - implying that focal absence of the body is due to an atrophic process. Since then, there has been increasing support for a theory that development occurs from multiple distinct foci, which later fuse. We describe a 3-year-old male with segmental agenesis of the corpus callosum, and provide neuroimaging in support of the latter theory. The patient is a 3-year-old male, born at 33 weeks. Pregnancy was complicated by maternal hypertension, BMI>50, and quarter pack-per-day cigarette use. The patient was admitted to the NICU with increased work of breathing, prolonged hypoglycemia, hypotension, and low free thyroxine with normal TSH. Further testing revealed an Insulin-like growth factor level of 37 ng/dL (15-150 ng/dL), a random growth hormone level of 0.91 ng/dL (<4.99 ng/dL), and a random cortisol level of 15.1 mcg/dL (3.0-22.0 mcg/dL). Testing excluded congenital adrenal hyperplasia. At an 8 week old evaluation with endocrinology, the patient passed an ACTH stimulation test, so further workup was not pursued. At 20 weeks, the patient underwent evaluation for episodic, non distractible, shuddering attacks, with an unremarkable EEG. At 45 weeks, the patient received a brainstem auditory evoked response test that was non-suggestive for hearing loss, despite concern on physical exam and at home. At 2 years and 11 months of age, the patient underwent evaluation for growth delay (length <3%, weight <10%). Insulin-like growth factor level was now 11 ng/dL (15-150 ng/dL.) Growth hormone response was tested via arginine/clonidine stimulation, and revealed severe growth hormone deficiency, prompting MRI evaluation of the pituitary gland. MRI of the brain revealed a Chiari I malformation (6mm displacement), a small pituitary gland (6.2 x 1.5 x 6.7mm) with infundibular thinning, and a complete absence of the callosal body despite

full development of the genu and splenium. This case further endorses the association of midline commissural defects with pituitary hypoplasia, adds support to the theory of multifocal, alinear, corpus callosum development, and displays the futility of non-gestationally adjusted, isolated, pituitary hormone levels.