it is unclear whether children with ACC present with early adaptive delays, or if difficulties emerge later as functional tasks become more complex. While potential delays in motor development are frequently reported, other domains such as communication, social and daily living skills are rarely described. We used a prospective, longitudinal design to examine adaptive behavior from 6-24 months in children with ACC and compared their trajectories to those with monogenic and neurodevelopmental conditions.

Participants and Methods: Our sample included children with primary ACC (n= 27-47 depending on time point) whose caregivers completed the Vineland Adaptive Behavior Scales-Interview 3rd Edition, via phone at 6, 12, 18 and 24 months. Comparison samples (using the Vineland-2) included children with Down Syndrome (DS; n = 15-56), Fragile X (FX; n = 15-20), children at high familial likelihood for autism (HL-; n=192-280), and low likelihood (LL; no family history of autism and no developmental/behavioral diagnosis; n = 111-196). A subset of the HL children received an autism diagnosis (HL+; n = 48-74). The DS group did not have an 18-month Vineland.

Results: A series of linear mixed model analyses (using maximum likelihood) for repeated measures was used to compare groups on three Vineland domains at 6, 12, 18 and 24 month timepoints). All fixed factors (diagnostic group, timepoint, and group X timepoint interaction) accounted for significant variance on all Vineland domains (p < .001). Post hoc comparisons with Bonferroni-correction examined ACC Vineland scores compared to the other diagnostic groups at each timepoint. At 6 months, parent-ratings indicated the ACC group had significantly weaker skills than the LL group in Communication and Motor domains. At 12, 18 and 24 months, ratings revealed weaker Communication, Daily Living and Motor skills in the ACC group compared to both the LL and HL- groups. Compared to the other clinical groups, the ACC group had stronger Socialization and Motor skills than Fragile X at 6 months, and at 24 months had stronger Communication and Socialization skills than both the DS and FX groups, as well as stronger Socialization than the HL+ group.

Conclusions: Compared to children with low likelihood of ASD, children with primary ACC reportedly have weaker Communication and Motor skills from 6 to 24 months, with weakness in Daily Living Skills appearing at 12 months and all differences increase with age. Compared to Fragile X, the ACC exhibited relative strengths in socialization and motor skills starting at 6 months. By 24 months, the ACC group was outperforming the monogenic groups on Socialization and Communication. In general, the ACC scores were consistent with the HL+ sample, except the ACC group had stronger Social skills at 18 and 24 months. The results clearly inform the need for early intervention in the domains of motor and language skills. Additionally, as we know that children with ACC are at increased risk for social difficulties, research is needed both using more fine-grained social-communication tools, and following children from infancy through middle childhood.

Categories: Behavioral Neurology/Cerebral Lateralization/Callosal Studies
Keyword 1: corpus callosum
Keyword 2: pediatric neuropsychology
Keyword 3: adaptive functioning
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2 Early Presentation of Autistic Features in Infants with Agenesis of the Corpus Callosum

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Objective: Agenesis of the Corpus Callosum (ACC), a congenital disorder in which the corpus callosum partially or completely fails to develop properly, occurs in ~1 in 4,000 live births. ACC has been identified as a major risk factor for receiving an Autism Spectrum Disorder (ASD) diagnosis (~10% diagnosis rate, as compared to 2.3% in the general population), but little is currently known about behavioral and social development during infancy and early childhood in this disorder. In this study we aim to 1) characterize the manifestation of autistic features in 24-month-old children (a common age for early diagnosis of ASD) with ACC and a convenience sample of age matched comparison children from the community, and 2) determine if parent reports of autistic features during the first year of life are associated with
reciprocal social behavior and autistic traits at 24-months of age in children with ACC.

Participants and Methods: Parents of infants who had received a diagnosis of ACC completed the Parent Rating of Communication, Emotion, and Social Skills (PROCESS) for their child at 6, 12, and 24-months of age, and the video-referenced rating of reciprocal social behavior (vrRSB) at 24-months of age. Comparison data were obtained from a convenience community sample in Minnesota.

Aim 1). The distributions of PROCESS total scores at 24-months were compared between the ACC group (n=43) and control group (n=1058) via a 2-tailed t-test. Likewise, distributions of reciprocal social behavior (RSB) scores at 24 months were compared between ACC (n=72) and controls (n=1438) via a 2-tailed t-test. Aim 2). A partial Pearson Correlation was conducted between ACC participants’ 6-month PROCESS scores and 24-month RSB (n=18) scores, as well as between their 12-month PROCESS and 24-month vrRSB (n=37) scores, controlling for child sex.

Results: Aim 1). At 24 months of age, children with ACC are reported to have significantly higher PROCESS scores ($t = 3.73, df = 42.67, p < .001$), and RSB ($t = 4.89, df = 88.38, p < .001$) scores than comparison toddlers, indicating an elevated presentation of behaviors associated with autistic features. Aim 2). No correlation was found between participants’ 6-month PROCESS and 24-month RSB scores ($r(16) = .39, p = .12$). A relatively strong correlation ($r(35) = .60, p < .001$) was found between participants’ 12-month PROCESS and 24-month RSB scores.

Conclusions: As early as two years of age, children with ACC display a heightened presentation of autistic features compared to typically developing controls. Additionally, reports of social behaviors related to ASD at 24-months are correlated with reports of autistic features at 12-months of age. This evidence indicates that children with ACC who are at a higher likelihood for being diagnosed with ASD may be identified as early as 12-months old. Formulating and leveraging an early identification methodology is imperative for this population with an already elevated risk for ASD, as providing early interventions leads to improved outcomes later in life.

Categories: Behavioral Neurology/Cerebral Lateralization/Callosal Studies

Keyword 1: corpus callosum

Keyword 2: autism spectrum disorder

Keyword 3: pediatric neuropsychology

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3 Emotional Expression in Infants with Agenesis of the Corpus Callosum: The Role of Callosal Connectivity in Early Temperament

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Objective: Accumulating evidence suggests that corpus callosum development is critically involved in the emergence of behavioral and cognitive skills during the first two years of life and that structural abnormalities of the corpus callosum are associated with a variety of neurodevelopmental disorders. Indeed by adulthood ~30% of individuals with agenesis of the corpus callosum (ACC), a congenital condition resulting in a partial or fully absent corpus callosum, exhibit phenotypic features consistent with autism spectrum disorder (ASD). However, very little is known about developmental similarities and/or differences among infants with ACC and infants who develop ASD. This study describes temperament in infants with ACC during the first year of life in comparison with a neurotypical control group. Additionally, it examines the potential contribution of disrupted callosal connectivity to early expression of temperament in ASD through comparison to children with high familial likelihood of ASD.

Participants and Methods: Longitudinal ratings of positive and negative emotionality were acquired at 6 and 12 months on the Infant Behavior Questionnaire–Revised across four groups of infants: isolated complete and partial ACC (n=104), high familial likelihood of ASD

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