Appendix D: Syndromes with constellations of features which overlap with FASD

Syndrome	Overlapping features	Features of this syndrome that differentiate it from FASD
Aarskog syndrome	Widely spaced eyes, small nose with anteverted nares, broad philtrum, mid-facial recession	Round face, down-slanted palpebral fissures, widow's peak, prominent "lop" ears, specific contracture of digits on extension. Inherited as an x-linked trait. Molecular defect identified.
Brachman-deLange or Cornelia deLange syndrome	Long philtrum, thin vermillion border of upper lip, depressed nasal bridge, anteverted nares, microcephaly	Single eyebrow across eyes and forehead (synophrys), long eyelashes, downturned corners of mouth, short upper limbs particularly involving ulnar side, very short stature. Molecular defect identified.
Dubowitz syndrome	Short palpebral fissures, widely spaced eyes, epicanthal folds, variable ptosis (droopy eyes) and blepharophimosis, microcephaly	Shallow supraorbital ridges, broad nasal tip, clinodactyly
Fetal anticonvulsant syndrome (includes fetal hydantoin and fetal valproate syndromes)	Widely spaced eyes, depressed nasal bridge, mid-facial recession, epicanthal folds, long philtrum, thin vermillion border of upper lip	Bowed upper lip, high forehead, small mouth
Maternal phenylketonuria (PKU) fetal effects	Epicanthal folds, short palpebral fissures, long poorly formed philtrum, thin vermillion border of upper lip, microcephaly	Prominent glabella, small upturned nose, round face
Noonan syndrome	Low nasal bridge, epicanthal folds, wide spaced eyes, long philtrum	Down-slanted palpebral fissures, wide mouth with well-formed philtrum, protruding upper lip. Molecular defect identified.
Toluene embryopathy	Short palpebral fissures, mid face hypoplasia, smooth philtrum, thin vermillion border upper lip, microcephaly	Large anterior fontanelle, hair patterning abnormalities, ear abnormalities
Williams syndrome	Short palpebral fissures, anteverted nares, board long philtrum, maxillary hypoplasia, depressed nasal bridge, epicanthic folds, microcephaly	Wide mouth with full lips and pouting lower lip, stellate pattern of iris, periorbital fullness, connective tissue dysplasia, specific cardiac defect of supravalvular aortic stenosis in many. Chromosome deletion on 7q (by chromosome microarray or specific 7q FISH (fluorescent in situ hybridization) probe analysis.
Other chromosome deletion and duplication syndromes	Many have short palpebral fissures, mid-facial hypoplasia, smooth philtrum	Chromosomal analysis by chromosome microarray

Adapted from: Chudley AE, Conry J, Cook JL, Loock C, Rosales T, LeBlanc N. Fetal Alcohol Spectrum Disorder: Canadian guidelines for diagnosis. *Can Med Assoc J*. 2005;**172**:S1-S21, (with permission of the author & journal)

Additional reference: Leibson T, Neuman G, Chudley AE, Koren G. The Differential Diagnosis of Fetal Alcohol Spectrum Disorder. *J Popul Ther Clin Pharmacol.* 2014; **21:** e1-30. https://jptcp.com/index.php/jptcp/article/view/347