Annex:

Reporting policy of fetal CNVs detected by aCGH

upon invasive prenatal testing.

prepared by the BeSHG Prenatal Committee on 14.09.2023

approved by the College for Medical Genetics on 02.02.2024

1. General guidelines

In general, specific aberrations are only reported when this is indicated by the BeSHG guidelines as found on the College for Genetics site:

*prenatal array

*susceptibility loci

2. List of possible aberrations to report or not to report

A non-exhaustive list of possible fetal aberrations can be found in the table below:

Aberration	Chromosome region	Genes	To report?	Literature / remarks
CMT1A **duplication** syndrome	17p12	*PMP22*	**YES**	Discussed with neurologists and presented on prenatal consortium meeting dd 17/09/2022.
22q11 **deletion** syndrome - **proximal**	22q11.21q11.22	LCR A-B or A-D	**YES**	Burnside et al; 2015
22q11 **deletion** syndrome - **central**	22q11.21q11.22	LCR B-D or C-D	**YES**	Burnside et al; 2015
22q11 **deletion** syndrome - **distal**	22q11.21q11.22	LCR C-E, D-E, D-F, E-F or E-H	**YES**	Burnside et al; 2015
22q11 **duplication** syndrome - **proximal**	22q11.21q11.22	LCR A-B or A-D	**YES**	Burnside et al; 2015
22q11 **duplication** syndrome - **central**	22q11.21q11.22	LCR B-D or C-D	**NO**	Burnside et al; 2015
22q11 **duplication** syndrome - **distal**	22q11.21q11.22	LCR C-E, D-E, D-F, E-F or E-H	**NO**	Burnside et al; 2015
STS **deletion**	Xp22.31	*STS*	**YES**	(discussed on prenatal consortium meeting dd 15/06/2023)
SHOX **duplication**	Xp22.33	*SHOX*	**NO**	

Aberration	Chromosome region	Genes	To report?	Literature / remarks
SHOX **deletion**	Xp22.33	*SHOX*	**YES**	
SHOX **deletion downstream**	Xp22.33	40kb region ~160kb downstream of *SHOX*: [hg19] Xp22.33(782334_822052)	**Clinical geneticist consult required**	Deletions and duplications upstream and downstream of the SHOX gene have been reported repeatedly as the cause for Leri-Weill Dyschondrosteosis and idiopathic small stature (Benito-Sanz et al., 2005 ; Chen et al., 2009 ; Bunyan et al., 2015). Clinical geneticist consult required: to report if relevant for phenotype (e.g. IUGR)

Version history

Version	Date prepared by BeSHG Prenatal Workgroup	Date approved by College of Medical Genetics	Updates
V2023	14.09.2023	02.02.2024	New document: no history available.