PRENATAL DIAGNOSIS VIA EXOME SEQUENCING

I. Prenatal diagnosis, via amniocentesis, CVS, or PUBS, using exome sequencing (81415, 81416, 81265, 88235) may be considered **medically necessary** when:

   A. The member's current pregnancy has either of the following:

      1. Non-immune hydrops fetalis, **OR**

      2. Two or more major malformations on ultrasound, which are affecting different organ systems, **AND**

   B. The member's current pregnancy has had a karyotype and/or microarray performed and the results were negative/normal, **AND**

   C. Alternate etiologies have been considered and ruled out when possible (examples: environmental exposure, injury, infection, maternal condition).

II. Prenatal diagnosis, via amniocentesis, CVS, or PUBS, using exome sequencing (81415, 81416, 81265, 88235) is considered **investigational** for all other indications.

III. Exome or genome sequencing (81265, 81415, 81416, 88235) for pregnancy loss on products of conception (POC) is considered **investigational**.

NOTES AND DEFINITIONS

1. **Major malformations** are structural defects that have a significant effect on function or appearance. They may be lethal or associated with possible survival with severe or moderate immediate or long-term morbidity. Examples by organ system include:
   - Genitourinary: renal agenesis (unilateral or bilateral), hypoplastic/cystic kidney

- Cardiovascular: complex heart malformations (such as pulmonary valve stenosis, tetralogy of fallot, transposition of the great arteries, coarctation of the aorta, hypoplastic left heart syndrome
- Musculoskeletal: osteochondrodysplasia/osteogenesis imperfecta, clubfoot, craniosynostosis
- Central nervous system: anencephaly, hydrocephalus, myelomeningocele
- Body wall: omphalocele/gastroschisis
- Respiratory: cystic adenomatoid lung malformation

REFERENCES

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2. Sparks TN, Lianoglou BR, Adami RR, et al. Exome Sequencing for Prenatal Diagnosis in Nonimmune Hydrops Fetalis [published online ahead of print, 2020 Oct 7]. N Engl J Med. 2020;10.1056/NEJMoa2023643. doi:10.1056/NEJMoa2023643